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Short Communication

Glossary of Medical and Molecular Genetics

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This French / English glossary of medical and molecular genetics is intended for students in human and biological sciences as well as medical and para-medical personnel. It is mainly a tool for teaching and research.
This glossary contains terminology frequently used in clinics and the laboratory. Within all areas of genetics the utilisation of terms in the glossary may also evolve with time or develop specific conations in different areas of study.
There is no direct correspondence between the French and English terms defined in these glossaries. Certain terms exist in only one of these languages. Also the utilisation of a given term may differ to some extent between French and English. The definitions of terms common to both glossaries are not necessarily literal translations of one another. Suggestions, corrections as well as the addition of new terms are welcomed.
We are grateful to the authors of those references who have contributed to the preparation of this glossary.

A

Acardia (French: acardia) Congenital absence of the heart.

Acellular system, see: system acellular.

Acentric (French: acentrique) Absence of centromere in a chromosome structure.

Achondrogenesis (French: achondrogénèse) Example of bone dysplasia due to a mutation in a collagen gene, COL2A1. Skeletal malformations are seen at ultrasound during the second trimester of pregnancy.

Acrocentric (French: acrocentrique) Position of the centromere near the end of a chromosome. Chromosomes 13-15 and 21,22 in man are acrocentric.

Adaptor (French: adapteur) Short nucleotidic sequence that has the property to link two DNA fragments that hat have no terminal complementary sequences.

Adenine (French: adénine) A nitrogenous base, one member of the base pair A-T, adenine- thymine.

ADN transfer, Southern blotting (French: marquage Southern) Transfer by absorption of DNA fragments separated in electrophoretic gels to membrane filters for detection of specific base sequences by radio-labelled complementary probes.

Advanced maternal age, AMA (French: âge maternel avancé, AMA) In most prenatal diagnostic clinics 35 years of age and more is considered as advanced maternal age due to the increased risk of chromosomal non disjunction in the foetus.

Affinity chromatography (French: chromatographie d'affinité) Any form of chromatography in which the components of the sample are separated on the basis of chemical affinity for a substance such as a binding protein or an immunoglobulin.

AFP, alpha foetoprotein (French: AFP, alphafoetoprotéine) Specific foetoglobulin synthesized by the liver and secreted in foetal serum during the foetal life and the neonatal period. An open spinal defect in the fetus is usually accompaniedby an increase in AFP in the amniotic fluid and a transudation towards the maternal circulation. AFP measurements in amniotic fluid and maternal serum are used in prenatal diagnosis of genetic diseases.

Agenesis (French: agénésie) Absence of an organ or structure.

(French: allèle) Alternative forms of a genetic locus; a single allele for each locus is inherited separately from each parent. Example: at a locus for eye colour the allele might result in blue or brown eyes.

Allelic frequencies (French: fréquences alléliques) Frequencies of allelic genes.

Allelomorph gene, see: alleles.

Allotransplant (French: allotransplant) Organ or tissue transplant between two individuals.

Alpha foetoprotein, see AFP.

Alu sequences (French: séquences ALU) DNA segments of approximately 300 base pairs with similar sequences. There are 50,000 copies in the human genome. They have a reconnaissance site for the ALU restriction enzyme.

AMA, see advanced maternal age.

Amino acid (French: acide aminé) Any of a class of 20 molecules that are combined to form proteins in living things. The sequence of amino acids in a protein and hence protein function are determined by the genetic code.

Amniocentesis (French: amniocentèse) Transabdominal or transuterine aspiration of amniotic fluid usually performed during the second trimester of pregnancy, for instance in the process of prenatal diagnosis of a genetic disease.

Amplifiable plasmid (French: plasmide amplifiable) Plasmid that continues to replicate even when host cell multiplication is blocked.

Amplification (French: amplification) An increase in the number of copies of a specific DNA fragment; can take place in vivo or in vitro. See cloning, polymerase chain reaction.

Amplification, DNA (French: amplification d'ADN) In vivo or in vitro increase in the number of a specific DNA fragments.

Amplification gene (French: amplification de gène) The increase in number of those genes needed for specialized functions in certain differentiated cells.

Analogue base (French: base analogue) Molecule which can replace a structure similar to nitrogenous DNA or RNA bases. Example : 5-bromo-uracil is a mutagenic analogue.

Anaphase (French: anaphase) Stage in and the first and second following the metaphase, during which the centromere splits and the chromatids which were lined up on the spindle begin to move apart towards opposite poles of the spindle.

Anencephaly (French: anencéphalie) Absence of cranial bone structure usually accompanied by a severe brain defect.

Aneuploidy (French: aneuploïdie) Situation when one or more chromosomes, missing or in excess of the normal modal number, is considered a deviation from the 2n ratio.

Angelman syndrome (French: Angelman, syndrome de) Angelman and Prader Willi syndromes are examples of syndromes related to parental imprinting. Both syndromes include mental retardation and clinical anomalies. They are due to the loss of a segment of chromosome 15 located in the proximal region of the long arm, 15q11q13. Depending on the parental origin of chromosome 15 and the exact location of the chromosomal anomaly, two different syndromes are identified.

Annealing (French: annelage) Hybridization of a synthetic oligonucleotide to a single strand nucleic acid. It is how a specific nucleotide sequence can be identified.

Anophtalmia (French: anophtalmie) Congenital absence of eye.

Anthelix (French: anthélix) Proximal fold of the external ear.

Antibody (French: anticorps) A specific substance produced by man, and animal, as a reaction to the presence of an antigen.

Anticipation (French: anticipation) Phenomenon in which the severity of the condition seems to increase or occur at an earlier age in subsequent generations.

Anticodon (French: anticodon) Group of three nucleotides located at one end of the transfer tRNA and by which it adapts, to the corresponding codon of the messenger mRNA, to fix the amino acid it carries.

Antigen (French: antigène) A substance which has the power of inducing, in man or in an animal, the formation of antibodies.

Antisense strand (French: brin anti-sens) Nucleic acid that has a sequence exactly opposite to a mRNA molecule made by the body; it binds to the mRNA molecule to prevent a protein from being made.

Apoptosis (French: apoptose) Programmed cell death.

Arachnodactyly (French: arachnodactylie) Long and thin finger(s).

Arhinencephaly (French: arhinencéphalie) Absence of mid-brain structure.

Arrayed library (French: banque de gènes) Individual primary recombinant clones (hosted in phage, cosmid, YAC, or other vector) that are placed in two-dimensional arrays inmicrotiter dishes. Each primary clone can be identified by the identity of the plate and the clone location (row and column) on that plate. Arrayed libraries of clones can be used for many applications, including screening for a specific gene or genomic region of interest as well as for physical mapping. Information gathered on individual clones from various genetic linkage and physical map analyses

is entered into a relational database and used to construct physical and genetic linkage maps simultaneously; clone identifiers serve to interrelate the multilevel maps.

Artificial gene (French: gène artificiel) A double-stranded DNA molecule, carrying a specific sequence, that will code for a specific amino acid sequence and that has been produced in vitro.

Ascites (French: ascite) Accumulation of fluid in the abdominal cavity.

Asymmetrical division (French: division asymétrique) Unequal segregation of chromosomes in mitosis. It leads to two daughter cells that have a different and abnormal number of chromosomes. Example: 46,XX leading to 47,XXX / 45,X.

A-T, see base pair.

AT, see ataxia telangiectasia.

Ataxia telangiectasia, AT (French: Ataxie télangiectasique, AT) Ataxia telangiectasia is an autosomal recessive disease characterized by a neurological and immunological symptomatology. Lymphoma and leukemia are often observed as complications of this syndrome. Cellular clones are identified that have chromosomal anomalies involving chromosomes 7 and 14. There is deficiency in repair of lesions induced by ionizing radiations.

Attenuator (French: atténuateur) A sequence of bases that occurs in the leader sequence of some operons and controls transcription. Synthesis of RNA may be terminated at this site.

Autogen regulation (French: régulation autogène) Regulation system where a gene product controls its own expression.

Autogenic recombination (French: rebombinaison autogène) Regulation system where a gene product controls its own expression.

Autolog graft (French: autogreffe) Cell or tissue graft derived from it own body.

Autoradiography (French: autoradiographie) A technique that uses X- ray film to visualize radioactively labelled molecules or fragments of molecules; used in analyzing length and number of DNA fragments after they are separated by gel electrophoresis.

Autosome (French: autosome) A chromosome not involved in sex determination. The diploid human genome consists of 46 chromosomes, 22 pairs of autosomes, and 1 pair of sex chromosomes : the X and Y chromosomes.

Auxotrophic (French: auxotrophie) Requiring a growth factor that is not required by the parental or prototype strain; may refer to microorganisms.

B

BAC, bacterial artificial chromosome (French: chromosome bactérien artificiel, BAC) A vector used to clone DNA fragments of 100 to 300 kb insert size, average of 150 kb in Escherichia coli cells. Based on the naturally occurring F-factor plasmid found in the bacterium Escherichia coli.

Back mutation (French: réversion vraie).

Bacteriophage (French: bactériophage), see phage.

Balanced translocation (French: translocation équilibrée) transposisiton of chromosomal segments without modification of the quantity of genetic material.

Banding, see chromosome banding.

Barr body (French: corpuscule de Barr), see sex chromatin.

Base pair (French: paire de bases) Two nitrogenous bases, adenine and thymine or guanine and cytosine, held together by weak bonds. Two strands of DNA are held together in the shape of a double helix by the bonds between base pairs.

Base pairing (French: appariement de bases) The pairing of nitrogenous bases in the polynucleotide chains by nitrogen bonds in a specific manner. The pairing occurs between a purine base of one strain and a pyrimidine of another strain in DNA, RNA or hybrid DNA/RNA molecules. In DNA the complementary bases pairs are Adenine and Thymine and Guanine and Cytosine. In RNA the base pairs are Adenine with Uracil and Guanine with Cytosine.

Base pair substitution (French: substitution d'une paire de bases) A class of lesions in DNA molecules which can give rise to gene mutations. They consist of transitions which preserve the purine-pyrimidine axis, and of transversions which reverse it.

Base ratio AT / GC (French: rapport de bases AT / GC) The amount of A equals the amount of T and the amount of G equals the amount of C but he amount of A+T does not equals the amount of G + C. This ratio is constant within a species but varies between species.

Base sequence (French: séquence de bases) The order of nucleotide bases in a DNA molecule.

Base sequence analysis (French: analyse d'une séquence de bases) A method, sometimes automated, for determining the base sequence.

Base triplets, triplet code, codon (French: triplets de bases) A sequence of 3 nucleotides comprising a codon of a nucleic acid and representing the code for an amino acid.

Bayesian analysis (French: analyse bayésienne) Mathematical method for calculating probality of the

carrier state in mendelian disorders combining several independent likelihoods.

Beckwith Wiedeman, syndrome (French: syndrome de Beckwith Wiedeman) Malformation syndrome characterized by a macroglossia, omphalocele and macrosomia. This is an example of a syndrome due to the phenomenon of parental imprinting. The gene is localized at 11p15.5.

Biotechnology (French: biotechnologie) A set of biological techniques developed through basic research and now applied to research and product development. In particular, the use by industry of recombinant DNA, cell fusion, and new bioprocessing techniques.

Bivalent (French: bivalent) A part of homologous chromosomes in association as seen at metaphase of the first meiotic division.

bp, see base pair.

Brachydactyly (French: brachydactylie) Short finger.

Brachyphalangy (French: brachyphalangie) Short phalanx.

BRCA1 / BRCA2 The first breast cancer genes to be identified. Mutated forms of these genes are believed to be responsible for about half the cases of inherited breast cancer, especially those that occur in younger women. Both are tumour suppressor genes.

Breakage syndrome (French: maladie cassante, syndrome d'instabilité chromosomique) Syndrome manifested by chromosomal anomalies, mainly chromosomal breakage and chromatid exchange.

Brushfield spots (French: taches de Brushfield) Presence of white speckles on the iris, a phenomenon commonly seen in Down syndrome.

C

CAAT sequence (French: CAAT, séquence) Sequence that has been conserved in some eukaryotic promoters. It lies about 40 base pairs upstream from the TATA box. This region controls the frequency of initiation by RNA polymerase.

Candidate gene (French: gène candidat) Gene suspected to be involved in the etiology of a disease.

Canthus (French: canthus) Inner and outer corners of the eyes.

CAP, catabolite activator protein (French: protéine CAP) The CAP is a positive regulatory protein, activated by cyclic AMP needed for RNA polymerase to initiate transcription of certain operons of E. coli. The CAP protein can bind at different sites relative to RNA polymerase.

Caruncle, see: canthus.

CAT assay (French: CAT, essai) Reporter gene assay used to measure activity of a promoter under different conditions, such as to define elements of a promoter or to study signals that activates an intact promoter.

CDNA, see: complementary DNA.

Cell colony (French: colonie cellulaire) Group of cells derived from the same initial cell.

Cell free system (French: système acellulaire) a preparation obtained from a broken cell preparation by differential centrifugation. A system composed of subcellular fractions and cell-free extracts, but devoid of intact cells.

Cell line (French: lignée cellulaire) A cultured cell type that can be reproduced indefinitely, or is immortalized.

Cell sorter (French: trieur de cellules) Apparatus that allows to sort different cell types or particles.

Cell strain (French: souche cellulaire) A population of animal cells that develops from a primary culture by reseeding serially; the characteristics of the parent cell are retained in culture.

Cell cycle (French: cycle cellulaire) One can distinguish 4 successive phases in a somatic cell life: mitosis, G1 phase, S phase of DNA replication and phase G2. Phases G1, S and G2 are found in interphase.

Centimorgan, cM (French: cM centimorgan) A unit of measure of recombination frequency. One centimorgan is equal to a 1% chance that a marker at one genetic locus will be separated from a marker at a second locus due to crossing over in a single generation. In human beings, 1 centimorgan is equivalent, on average, to 1 million base pairs.

Centric fusion (French: fusion centrique) Fusion of chromosomes at the level of the centromere.

Centromere, kinetochore (French: centromère) A specialized chromosome region to which spindle fibres attach during cell division.

Cephalometry (French: céphalométrie) Measurement of the head.

Chiasma (French: chiasma) Exchange sites between chromatids observed in prophase of meiosis 1 subsequent to crossing over.

Chimera (French: chimera) An organism that contains cells or tissues with a different phenotype. These can be mutated cells of the host organism or cells from a different organism or species.

Chimeric gene, hybrid gene (French: gène chimérique) Gene made of DNA fragments of different origins.

Chorionic villus sampling, chorionic villi (French: villosités choriales, prélèvement de) A procedure used for prenatal diagnosis at the end of the first trimester around the 12th to the 14th week of pregnancy. Fetal tissue is withdrawn from the villus area of the chorion either trans uterine cervix or trans abdominal under ultrasonic guidance.

Chromatid (French: chromatide) A duplicated chromosome is formed by two longitudinal units or chromatids joined at the centromeric region. A chromosome is formed by two sister chromatids. Each

chromatid is made of a long DNA strand unique and identical to the sister chromatid but different from the homologue chromosome originating from the other parent.

Chromatid exchange, see: crossing over.

Chromatin (French: chromatine) Constitutive structure of chromosomes made of DNA, basic chromosomal proteins and small RNA quantities. Chromatin is visible during interphase.

Chromonema (French: chromonéma, chromonème) The thin, threadlike form of an uncoiled chromosome seen in interphase, early prophase, and late telophase, as opposed to the tightly coiled metaphase chromosome.

Chromosomal anomaly (French: anomalie chromosomique) A chromosome anomaly can be constitutional, acquired, homogenous, mosaic, numerical, structural.

Chromosomal condensation (French: condensation chromosomique) Induction of chromosomal condensation in an interphase nucleus by fusion with a cell in mitosis. Condense chromosomes in S phase appear pulverized.

Chromosome (French: chromosome) The self-replicating genetic structures of cells containing the cellular DNA that bears in its nucleotide sequence the linear array of genes. In prokaryotes, chromosomal DNA is circular, and the entire genome is carried on one chromosome. Eukaryotic genomes consist of a number of chromosomes whose DNA is associated with different kinds of proteins.

Chromosome banding (French: bande chromosomique) Chromosome regions identified by specific stains.

Chromosome instability syndrome (French: syndrome d'instabilité chromosomique) Genetic disease characterized by an increase rate of chromosomal breaks, exchange between sister chromatids and spontaneous structural anomalies.

Chromosome jumping (French: saut chromosomique) Methods for obtaining at a considerable distance from an initial cloned fragment without the need for overlapping clones of DNA regions between the two sites.

Chromosome puff (French: puff chromosomique) An enlarged region along a polytene chromosome, the site of active transcription.

Chromosome set (French: haplome) See genome.

Chromosome walking (French: marche le long d'un chromosome) Sequential isolation of molecular clones in order to span large intervals on the chromosome.

Cis control (French: contrôle en cis).

Cis-trans position effect (French: effet de position cis-trans) Terms cis and trans describe the gene position on homologue chromosomes in double heterozygote individuals. When two alleles are located near each other on the same chromosome,they are in cis position. When they are located on different homologue chromosomes they are in trans position. Cis and trans are analogue to coupling and repulsion.

Cistron (French: cistron) Genetic function unit defined by the cis-trans test. The smallest unit of genetic material that must be intact to function as a transmitter of genetic information.

Cleavage site (French: site de coupure)The cleavages generated by restriction endonucleases can be within or immediately adjacent to the recognition sequence but in some cases the cleavages are displaced from the recognition sequence by a specific number of nucleotides.

Cleft palate (French: fente palatine) Fissure of the mucous membrane and / or the bone structure of the palate.

Clinical heterogeneity (French: hétérogénéité clinique) Different phenotypes due to mutations in the same gene.

Clinodactyly (French: clinodactylie) Incurved finger.

Clone (French: clone) A group of cells derived from a single ancestor.

Clone bank (French: banque de clones) See genomic library.

Clone sorting (French: criblage) Operation of identification and sorting of clones.

Cloning (French: clonage) The process of asexually producing a group of cells (clones), all genetically identical, from a single ancestor. In recombinant DNA technology, the use of DNA manipulation procedures to produce multiple copies of a single gene or segment of DNA is referred to as cloning DNA.

Cloning vector (French: vecteur de clonage) DNA molecule originating from a virus, a plasmid, or the cell of a higher organism into which another DNA fragment of appropriate size can be integrated without loss of the vectors capacity for self-replication; vectors introduce foreign DNA into host cells, where it can be reproduced in large quantities. Examples are plasmids, cosmids, and yeast artificial chromosomes; vectors are often recombinant molecules containing DNA sequences from several sources.

Club foot (French: pied bot) Congenital deformation of the foot.

cM, see centimorgan.

Coarctation of aorta (French: coarctation de l'aorte) Congenital cardiac anomaly manifested by a constriction of the aorta isthmus.

Code, see : genetic code.

Coding sequence, see: sequence, coding.

Codominant (French: codominant) Two alleles that are expressed simultaneously in the heterozygote state. Example the ABO blood group.

Codon, see: genetic code.

Coefficient of inbreeding, see: parental coefficient.

Coloboma (French: colobome) Congenital defect seen as a fissure. Examples: coloboma of the eyelid, iris, retina…

Compatibility (French: compatibilité) Tissular compatibility or histocompatibility allows successful grafts; histocompatibility of the donor and recipient are identical or very close.

Complementary DNA, cDNA (French: ADN complémentaire, ADNc) DNA that is synthesized from a messenger RNA template; the single-stranded form is often used as a probe in physical mapping.

Complementary sequences (French: séquences complémentaires) Nucleic acid base sequences that can form a double- stranded structure by matching base pairs; the complementary sequence to G- T- A- C is C- A- T- G.

Complementation (French: complementation) Complementary effect of a double mutation on different genes. The two mutant genes may reconstitute a normal phenotype. Xeroderma pigmentosum is an example of complementation.

Compound heterozygote, see heterozygote composite.

Conditional mutation (French: mutation conditionnelle) Mutation expressed only under certain conditions. Lethal mutations exist in a cell but in the homozygote state under conditional forms, like for instance if they are expressed at certain temperatures.

Confined placental mosaicism (French: mosaïque placentaire) Chromosomal mosaicism limited to placental tissues.

Congenital (French: congénital) Presence at birth of a trait or defect.

Conjugation, mating (French: conjugaison) Natural transfer of plasmidic or chromosomal DNA from a bacterial cell to another via a cytoplasmic bridge.

Consanguinity coefficient (French: coefficient de consanguinité) Probality for an individual to carry two copies of the same gene transmitted by his parents who have a common ancestor.

Conserved sequence (French: séquence conservée) A base sequence in a DNA molecule (or an amino acid sequence in a protein) that has remained essentially unchanged throughout evolution.

Constitutive gene (French: gene constitutive) Gene expressed without particular regulation.

Constitutive mutation (French: mutation constitutive) Mutation that inhibits the normal gene regulation; its expression becomes independent of environmental factors.

Constriction (French: constriction) Reduced thickness in a chromosomal region. The centromere is the primary constriction. Chromosome 1,9,16 have a secondary constriction.

Contact inhibition (French: inhibition de contact) The inhibition of cell division and cell motility in normal animal cells when in close contact with each other.

Contig map (French: carte de contig) A map depicting the relative order of a linked library of small overlapping clones representing a complete chromosomal segment.

Contigs (French: contigs) Group of clones representing overlapping regions of a genome used to determine the physical map of a chromosome region. Contig is the abbreviation for contiguous.

Copy error (French: erreur de copie) an error that arises during DNA replication from a failure to insert nucleotides complementary to those in the parent DNA chain.

Corepressor (French: corépresseur) Affector molecule that modifies a regulatory protein (activation), allowing it to bind to an operator to inactivate transcription.

Cosmid (French: cosmide) Artificially constructed cloning vector containing the cos gene of phage lambda. Cosmids can be packaged in lambda phage particles for infection into E. coli; this permits cloning of larger DNA fragments (up to 45 kb) than can be introduced into bacterial hosts in plasmid vectors.

Coupling (French: couplage) The occurrence on the same chromosome in a double heterozygote of the two mutant genes of interest, the normal alleles being on the homologous chromosome. The genes are said to be linked in coupling.

Craniolacuna (French: craniolacunie) Circumscribed deficient cranial bone defect.

Craniosynostosis (French: craniosynostose) Fusion of cranial sutures.

Crossing over (French: enjambement) The breaking during meiosis of one maternal and one paternal chromosomes, the exchange of corresponding sections of DNA, and the rejoining of the chromosomes. This process can result in an exchange of alleles between chromosomes.

Cybrid (French: cybride) Cytoplasmic hybrid resulting from the fusion of protoplasts. A hybrid contains the cytoplasmic and nuclear genetic information from the two parental cells.

Cycline (French: cycline) Protein family that plays an important role in the regulation of cell division.

Cyst (French: kyste) Abnormal closed cavity, of various sizes in which there is a liquid collection of infectious or embryological origin.

Cystic fibrosis, mucoviscidosis (French: fibrose kystique, mucoviscidose) A disease affecting the pancreas, gastro intestinal and pulmonary functions and due to several different mutations in the CFTR gene located on chromosome 7 at the 7q31.2 locus.

Cytoplasmic gene (French: gène extrachromosomique) Any gene that ordinarily exists on nucleic acid in the cytoplasm, especially on mitochondrial or chloroplast chromosomes.

Cytoplasmic inheritance (French: hérédité cytoplasmique, maternelle, extranucléaire) The inheritance of traits controlled by genes located on the DNA of mitochondria.

Cytosine-C (French: cytosine-C) A nitrogenous base, one member of the base pair G- C (guanine and cytosine).

D

Dalton atomic mass unit (French: Dalton, unité de masse) Atomic mass unit. One Dalton corresponds to one hydrogen atom (1,657 x 10 –24).

DAPI (French: DAPI) Chromosome stain derived from fluorescent stain <4',6-diamidino-2-phenylindole> that stains preferentially heterochromatin of chromosomes 9,15 and Y.

Defective prophage, see prophage, defective.

Deformation (French: déformation) Malformation due to an abnormal position of a limb or an abnormal pressure.

Dehiscence (French: déhiscence) The formation of a fissure.

Deletion (French: délétion) Loss of part of a whole chromosome or loss of DNA nucleotide bases.

Denaturation (French: dénaturation) Formation of a single DNA strand from a double strain under heating of chemical bonds responsible for base pairing.

Denaturation mapping (French: cartographie par dénaturation partielle) A method employing electron microscopy of DNA partially denaturated which permits the decision as to whether the gene sequence in a viral genome is linear or circularly permuted.

De novo mutation (French: mutation de novo, néo-mutation) Spontaneously occurring mutation.

Deoxyribonucleotide, see nucleotide.

Derepression (French: dérépression) An increase in the synthesis of the product of a regulated gene by interference with the action of a repressor. It can be produced by the mutation of the repressor gene or of the operator gene or by an inducer that binds to the repressor, releasing it from the operator.

Dermatoglyphics (French: dermatoglyphes) The study of the surface markings of the skin. Dermatoglyphic studies are used in a number of malformation syndromes due to a chromosomal aberration as in trisomy 21 or Down syndrome.

Diakinesis (French: diacinèse) Terminal phase of the first meiotic division during which the chromosomes are tightly bond and strongly coloured by the staining solutions.

Dicentric (French: dicentrique) An aberrant chromosome that contains two centromeres.

Differentiation (French: différenciation) The act or process of acquiring completely individual characters, such as occurs in the progressive diversification of cells and tissues of the embryo.

DiGeorge, Velo Cardio Facial, CATCH 22, syndrome (French: DiGeorge, VeloCardio Facial, CATCH 22, syndrome) Syndrome due to a deletion at the 22q11.2 locus, involving several genes accompanied by, in VCF, facial dysmorphism, palatal insufficiency, heart defect or, in Di George syndrome, parathyroid hypoplasia, thymus hypoplasia, and outflow of the heart defect.

Diploid (French: diploïde) A full set of genetic material, consisting of paired chromosomes one chromosome from each parental set. Most animal cells except the gametes have a diploid set of chromosomes. The diploid human genome has 46 chromosomes.

Diplotene (French: diplotène) One prophase stage of the first meiotic division.

Discordant (French: discordant) A twin pair (or set of individuals) in which one member exhibits a certain trait and the other does not.

Disomy uniparental, UPD, see uniparental disomy.

Dispermia (French: diandrie) Presence of two spermatozoid during the fertilization likely to lead to a triploidy.

Disruption (French: disruption) In the disruption sequence the fetus is subject to a destructive problem. It may be a vascular, infectious or mechanical problem leading to a malformation. One example is the effect of an amniotic band.

Dizygote (French: dizygotie) The product of fertilization of two separate eggs by two separate sperms; non identical twin pair.

DNA, deoxyribonucleic acid (French: ADN, acide désoxyribonucléique) The molecule that encodes genetic information. DNA is a double- base pairs of nucleotides. The four nucleotides in DNA contain the bases stranded molecule held together by weak bonds between base pairs of nucleotides. The four nucleotides in DNA contain the bases: adenine (A), guanine (G), cytosine (C), and thymine (T). In nature, base pairs

form only between A and T and between G and C; thus the base sequence of each single strand can be deduced from that of its partner.

DNA probes, see: probe.

DNA repair (French: réparation de l'ADN) Genes encoding proteins that correct errors in DNA sequencing.

DNA replication (French: replication d'ADN) The use of existing DNA as a template for the synthesis of new DNA strands. In humans and other eukaryotes, replication occurs in the cell nucleus.

DNA-RNA hybrid (French: hybride ADN-ARN) A double helix consisting of one chain of DNA hydrogen bonded to a complementary chain of RNA.

DNA sequence (French: séquence d'ADN) The relative order of base pairs, whether in a fragment of DNA, a gene, a chromosome, or an entire genome. See: base sequence analysis.

DNA synthetizer (French: synthétiseur d'ADN) An automated machine design to synthesize short polynucleotide chains -oligonucleotides- similar in structure to DNA.

DNA transfer, see: Southern blotting.

Domain (French: domaine) Discrete portion of a protein with its own function. The combination of domains in a single protein determines its overall function.

Dominant (French: dominant) An allele that is almost always expressed, even if only one copy is present.

Double helix (French: double hélice) The shape that two linear strands of DNA assume when bonded together.

Double heterozygote, see: composite, compound heterozygote.

Double minute chromosomes (French: chromosome minuscule double) Fragment of unstable extrachromosomic DNA without a centromere. Minute chromosomes are small spherical structures, observed in pairs in varying number from cell to cell. They stain homogeneously. They are duplicated once in mitosis and are lost in cellular division.

Ductus arteriosus, patent (French: persistance du canal artériel) Persistence of the arterial canal after birth.

Duplicate (French: dupliquer) Make a copy.

Dysgenic (French: dysgénique) Detrimental to the hereditary qualities of man or tending to counteract racial improvement through an influence bearing on reproduction. Refers to a genetic anomaly.

Dysmorphism (French: dysmorphisme) Developmental anomaliy seen in a variety of syndromes with a genetic or environmental origin.

Dysostosis (French: dysostose) Defective ossification.

Dysplasia (French: dysplasia) Abnormality of development.

Dysraphia (French: dysraphie) Developmental defect of the median raphe. Examples: spina bifida, cleft lip.

Dysraphism (French: dysraphisme) Term used to describe a neural tube or spine anomaly.

E

E. coli (French: E coli) Escherichia colli is a common bacterium that has been studied intensively by geneticists because of its small genome size, normal lack of pathogenicity, and ease of growth in the laboratory.

Ectoderm (French: ectoderme) The outer of three layers of cells comprising the early embryo. Gives rise to skin and neural tissue.

Effusion (French: épanchement) Abnormal presence of fluid in a tissue or cavity.

Electrofocusing, electro focalisation, see isoelectric focusing.

Electrophoresis (French: électrophorèse) A method of separating large molecules (such as DNA fragments or proteins) from a mixture of similar molecules. An electric current is passed through a medium containing the mixture, and each kind of molecule travels through the medium at a different rate, depending on its electrical charge and size. Separation is based on these differences. Agarose and acrylamide gels are the media commonly used for electrophoresis of proteins and nucleic acids.

Electroporation (French: électroporation) Technique used to facilitate the penetration of DNA in cells based on the use of electric pulsions to increase the membrane permeability.

Embryo (French: embryon) fetus before the end of the third month.

Embryonic biopsy (French: biopsie embryonnaire) Biopsy of embryonic tissues usually performed on spontaneous miscarriage material.

Embryonic stem cell, see: stem cell.

Encephalocele (French: encéphalocèle) Cranial defect of the midline with hernia of the meningeal membrane containing spinal fluid and abnormal brain tissue.

3' End, terminus (French: extrémité 3') The end of a polynucleotide chain terminating with a 3' carbon atom. The 5' position of one pentose ring is connected to the 3' position of the next pentose ring via a phosphate group. All RNA chains, as well as DNA chains, grow in the 5' to 3' direction.

5' End, terminus (French: extrémité 5') The beginning of a polynucleotide chain terminating which has a free 5' group. The 5' position of one pentose ring is connected to the 3' position of the next pentose ring via a phosphate group. All RNA chains, as well as DNA chains, grow in the 5' to 3' direction.

Endoderm (French: endoderme) One of three layers of cells comprising the early embryo It gives rise to the lining of the gut and cells of the pancreas and liver.

Endonuclease (French: endonucléase) An enzyme that cleaves its nucleic acid substrate at internal sites in the nucleotide sequence.

Endoreduplication (French: endoreduplication) Presence of duplicated chromosomes in metaphase with four chromatids and two centromeres.This duplication is total if all chromosomes are concerned and selective if only one chromosome or part of it is duplicated.

Enhancer (French: amplificateur) Element that increases the utilization of (some) eucaryotic promoters in cis configuration, but that can function in any 1ocation, upstream or downstream, relative to the promoter.

Enzyme (French: enzyme) A protein that acts as a catalyst, speeding the rate at which a biochemical reaction proceeds but not altering the direction or nature of the reaction.

Epicanthus (French: épicanthus) A fold of skin projected over the inner canthus.

Epidemiology (French: épidémiologie) Study of the different factors that intervene in the onset and evolution of diseases.

Epigenetic (French: épigénétique) The term that refers to any factor that can affect the phenotype without change in the genotype.

Equatorial plane (French: plan équatorial) Alignment of chromosomes at metaphase in a ring formation at the nuclear circumference.

EST, Expressed sequence tag, see sequence tagged site.

Euchromatin (French: euchromatine) Region of chromosomes that have a normal cycle of spiralisation-despiralisation by opposition to heterochromatin.

Eukaryote (French: eucaryote) Cell or organism with membrane-bound, structurally discrete nucleus and other well- developed subcellular compartments. Eukaryotes include all organisms except viruses, bacteria, and blue- green algae.

Euploidy (French: euploïdie) Normal number of chromosomes.

Evolutionarily conserved, see: conserved sequence.

Exogenous DNA (French: ADN exogène) DNA originating outside an organism.

Exons (French: exon) The protein-coding DNA sequences of a gene.

Exonuclease (French: exonucléase) An enzyme that cleaves nucleotides sequentially from free ends of a linear nucleic acid substrate.

Expressed gene, see gene expression.

Expressivity (French: expressivité) Intensity of gene expression from mild to severe.

Extremity 3', see: 3' end terminus.

Extremity 5', see: 5' end terminus.

F

Fallot tetralogy (French: tétralogie de Fallot) Congenital heart defect manifested by an inversion of the great vessels, and a ventricular septal defect.

Familial (French: familial) Relevant to a trait that is more frequent in members of an affected individual family than in the general population. This may be caused by genetic or environmental factors, or both.

Family history (French: histoire familiale) Family history based on genealogy to identify individuals carriers of or affected with a trait or genetic disease similar to the propositus genotype or phenotype.

Family tree (French: pedigree, histoire familiale) Ancestral information in relation to an individual or family.

Fibroblast (French: fibroblaste) Young conjonctive cell.

Filling in (French: remplissage) Insertion of nucleotides in a single DNA strand to make it entirely double stranded.

Finger loop (French: boucle digitale) Image formed by the epidermal ridges on finger tips. Radial and cubital loops are found as well as whorls and arches. One notes an excess of arches on the fingertips in trisomy 18.

Finger printing (French: cartographie peptidique) Analytical method that supplies an exact identity card of a protein molecule containing electrophoretic and chromatographic properties of various polypeptide segments of the molecule.

FISH Fluorescent in situ hybrization (French: hybridation in situ en fluorescence) A physical mapping approach that uses fluorescein tags to detect hybridization of probes with metaphase chromosomes and with the less- condensed somatic interphase chromatin.

Flow cytometry (French: cytométrie de flux)Analysis of biological material by detection of the light-absorbing or fluorescing properties of cells or subcellular fractions (i.e., chromosomes) passing in a narrow stream through a laser beam. An absorbance or fluorescence profile of the sample is produced.

Automated sorting devices, used to fractionate samples, sort successive droplets of the analyzed stream into different fractions depending on the fluorescence emitted by each droplet.

Flow karyotyping (French: caryotypage de flux) Use of flow cytometry to analyze and/or separate chromosomes on the basis of their DNA content.

Fondamental research (French: recherche fondamentale) Research that studies normal and abnormal physiological processes, while clinical

research is applied to prevention, diagnostic and cure of disease.

Founder effect (French: effet fondateur) The high frequency of a mutant gene in a rapidly expanding population founded by a small ancestral group when one or more of the founders were, by chance, carriers of the mutant gene.

Fragile site (French: site fragile) Non-staining gaps, known as fragile sites, are occasionally observed in characteristic sites on several chromosomes.

Fragile X (French: X-fragile) Common form of X linked mental retardation associated with a fragile site on X chromosome. The genetic defect has been identified as an abnormal amplification or more than 60 times of triple CGG at the Xq27.3 locus. If the child has the disease the mother can have a pre-mutation with an abnormal amplification between 60-200 repeats of the CGG triplet. The amplification hampers the expression of gene FMR-1. FRAXA site is not far from FRAXE site, a variant of the syndrome.

Fragment (French: fragment) Part of a chromosome detached by breakage : if there is a centromere we refer to a centric fragment and if not it is called acentric.

Frameshift (French: décalage du cadre de lecture) A shift in the reading frame used to translate the base sequence of mRNA. It is caused by the addition or deletion of one or more bases, resulting in an alternative peptide being formed.

Frameshift mutation (French: mutation de changement de phase) Change resulting from addition or deletion of nucleotides, in numbers other than three (French : triplets; codons), which moves the translation 'reading frame' so that a new set of codons, beyond the point of abnormality in the messenger RNA, is read.

FRAXA, FRAXE, see: fragile X

G

Gamete (French: gamète) Mature male or female reproductive cell, sperm or ovum, with a haploid set of chromosomes or 23 for humans.

G-C, see: base pair.

Gemellity, twinning (French: gémellité) Simultaneous development of two embryos. It is univitelline if originating from an egg subdivision or bivitelline if several eggs were fertilized at the same time.

Gene (French: gène) The fundamental physical and functional unit of heredity. A gene is an ordered sequence of nucleotides located in a particular position on a particular chromosome that encodes a specific functional product: a protein or RNA molecule.

Gene amplification, see amplification gene.

Gene candidate (French: gène candidat) Gene suspected to be responsible for the occurrence of a genetic disease.

Gene cluster (French: groupe de gènes) Any group of two or more closely linked genes on a chromosome that are related functionally.

Gene construction (French: Construction génique) Portion of DNA involved in the transfer into a cell containing an interest gene and the promoter and regulator sequences essential to its expression and regulation in the receptor cell.

Gene conversion (French: conversion de gène) Unequal recovery of genetic markers or alleles in the region of the exchange during genetic recombination. Processes analogous to meiotic gene conversion in eukaryotes. Changes from heterozygosity to homozygosity by mechanism other than segregation.

Gene expression (French: expression génique) The process by which a gene coded information is converted into the structures present and operating in the cell. Expressed genes include those that are transcribed into mRNA and then translated into protein and those that are transcribed into RNA but not translated into protein.

Gene families (French: familles de gènes) Groups of closely related genes that make similar products.

Gene flow (French: flux génétique) The spread of genes from one breeding population to others owing to the dispersal of gametes or zygotes.

Gene fusion (French: fusion de gènes) Association of gene fragments leading to the formation of a chimera.

Gene library, see genomic library.

Gene mapping (French: cartographie génique) Determination of the relative positions of genes on a DNA molecule, chromosome or plasmid, and of the distance, in linkage units or physical units, between them.

Gene product (French: produit génique) The biochemical material, either RNA or protein, resulting from expression of a gene. The amount of gene product is used to measure how active a gene is; abnormal amounts can be correlated with disease- causing alleles.

Gene rearrangement (French: réarrangement génétique) Collection of several pieces of DNA initially not contiguous.

Gene tagging (French: étiquetage génétique) Insertion of a genetic marker in or close to a gene.

Gene therapy (French: thérapie génique) Insertion of normal DNA directly into cells to correct a genetic defect.

Gene transfer (French: transfert de gène) Transfer of genetic material from one cell to another or from one organism to another.

Genetic code (French: code génétique) The sequence of nucleotides, coded in triplets (codons) along the mRNA, that determines the sequence of amino acids in protein synthesis. The DNA sequence of a gene can be used to predict the mRNA sequence, and the genetic

code can in turn be used to predict the amino acid sequence.

Genetic competence (French: compétence génétique) Cellular state that allows penetration of a foreign nucleic acid.

Genetic complementation, see complementation.

Genetic consensus (French: consensus génétique) Short DNA sequence found in a number of genes and living organisms.

Genetic construction (French: construction génique) Portion of DNA needed for the transfer in a cell of a gene of interest including the promoter and regulators essential to its expression and regulation in the receiving cell.

Genetic conversion, gene conversion (French: conversion génique) Non reciprocal genetic recombination. Interaction between allelic sequences during meiosis leading to an unequal exchange of genetic information.

Genetic diagnosis (French: diagnostic génétique) Detection of genes of an organism by hybridization of its genome with specific molecular probes. Diagnosis of a genetic disease.

Genetic disease (French: maladie génétique) Disease due to the mutation of one or several genes. When only one gene is involved, we refer to a monogenic disease.

Genetic disruption (French: disruption génique) Interruption of the coding sequence of a gene due to the introduction of another DNA sequence.

Genetic distance (French: distance génétique) Degree of filiation between different genomes. In molecular genetics the genetic distance is measured by the percentage of structural homology between two sequences by molecular hybridization or by comparing nucleotide sequences.

Genetic drift (French: dérive génétique) Any change either directed or undirected in gene frequency in a population.

Genetic engineering (French: génie génétique) Manipulations by which an individual having a new combination of inherited properties is established.

Genetic footprint (French: empreinte génétique) Fine structural characteristic of a specific DNA region allowing to identify a specific cell and its association.

Genetic heterogeneity (French: Hétérogénéité génétique) Presence of similar phenotypes due to different genetic mechanisms.

Genetic load (French: fardeau génétique) In humans we refer to hereditary defects which lower life expectancy or reduce reproduction capacity.

Genetic map, see linkage map.

Genetic material (French: matériel génétique) See : genome.

Genetic polymorphism (French: polymorphisme génétique) The regular and simultaneous occurrence in the same population of two or more alleles at a genetic locus, with at least one minor allele having a frequency greater than 1%.

Genetically modified organism, GMO (French: organisme génétiquement modifié, OGM) Organism of which the genome has been modified. If the reproductive cells carry the modification they will transmit it to the descents.

Genetics (French: génétique) The study of the patterns of inheritance of specific traits.

Genic conversion, see: genetic conversion.

Genocopy (French: génocopie) The production of the same phenotype by different genes also called "mimetic genes".

Genome (French: génome) All the genetic material in the chromosomes of a particular organism; its size is generally given as its total number of base pairs.

Genome map (French: carte génomique) Research and technology development efforts aimed at mapping and sequencing some or all of the genome of human beings and other organisms.

Genome sequencing (French: séquençage du génome) Determination of the order in which the bases are arranged within a length of DNA or RNA or, the sequence of amino acids that make up a protein.

Genomic library, gene bank (French: banque de gènes) A collection of clones made from a set of randomly generated overlapping DNA fragments representing the entire genome of an organism.

Genotype, haplome (French: génotype) The sum of genetic information or gene contained in the chromosomes of the individual as distinguished from their phenotype. It determines not a unique phenotype but a range of phenotype capacities referred to as an individual's "norm of reaction" to the environment.

Germinal stem cell, see stem cell.

Glabella (French: glabelle) Region of the face between eyebrows.

GMO, see: modified organism.

Guanine-G (French: guanine-G) A nitrogenous base, one member of the base pair G-C or guanine and cytosine).

H

Haploid (French: haploïde) A single set of chromosomes: half the full set of genetic material present in the egg and sperm cells of animals and in the egg and pollen cells of plants. Human beings have 23 chromosomes in their reproductive cells.

Haplome, see: genotype.

Haplotype (French: haplotype) Genetic material carried by only one of two chromosomes and corresponding to a specific character.

Harelip (French: fente labiale) Uni or bilateral closure defect of the upper lip.

Hare lip and cleft palate (French: gueule de loup, fente labio-palatine) Hare lip and cleft palate frequently observed in holoprosencephaly or a chromosomal defect like the trisomy 13.

HAT culture medium (French: milieu de culture HAT) A tissue culture medium containing hypoxanthine, aminopterin anbd thymidine, used in somatic cells fusion experiments.

HDL, high density cholesterol (French: cholestérol de haute densité, HDL) Cholesterol transported by high density lipoproteins.

Helix (French: helix) The margin of the external ear.

Hematopoietic (French: hématopoïétique) Related to the formation of blood cells, a process that occurs mainly in the bone marrow.

Hemizygosity (French: hémizygotie) Presence of a single gene copy in a diploid cell, example : X and Y in the male. In abnormal situations there may be a deletion on a chromosome or entire loss of that chromosome leading to a partial or complete deletion, ex Turner 45X syndrome or the Cri du Chat 5p- anomaly.

Heritability (French: héritabilité) That portion of the character variance due to hereditary factors as distinct from factors of environment.

Heterochromatin (French: hétérochromatine) A fraction of chromatin that has characteristics distinct from the bulk of the chromosomal material, the euchromatin.

Heteroduplex (French: hétéroduplex) A DNA double helix in which the complementary strands are derived from different molecules, so that there may be small regions of mismatching.

Heteroduplex technique, mapping (French: technique d'hétéroduplex) Technique that allows to visualize in electron microscopy non homologous segments of a mutant from the normal type.

Heterokaryon (French: hétérocaryon) A cell in which genetically different haploid nuclei may co-exist and multiply.

Heteromorphism (French: hétéromorphisme) Normal morphological variation in chromosome staining.

Heteroplasmy (French: hétéroplasmie) Presence of more than one type of mitochondria in cells of an individual : simultaneous presence of normal DNA and mutant mtDNA.

Heteroploidy (French: hétéroploïdie) Any different chromosome number from normal.

Heterosis (French: hétérosis) Advantage of heterozygote genotypes over homozygote genotypes of their parents.

Heterozygosity (French: hétérozygosité) The presence of different alleles at one or more loci on homologous chromosomes.

Heterozygote composite, compound (French: hétérozygote composite) Individual who is heterozygote at two different loci.

Hexadactylia (French: hexadactylie) Autosomal dominant malformation characterized by the presence of a sixth digit on hands and or feet.

Histone (French : histone) Basic chromosomal proteins present in eukaryotes believed to be involved in the coiling and condensation of chromosomes.

Holandric (French: holandrique) Hereditary trait due to the presence of a gene on Y chromosome. Very few characters are linked to the Y chromosome. The transmission of the trait is exclusively from father to son.

Homeobox (French: homéobox) A short stretch of nucleotides whose base sequence is virtually identical in all the genes that contain it. It has been found in many organisms from fruit flies to human beings. In the fruit fly, a homeobox appears to determine when particular groups of genes are expressed during development.

Homoduplex (French: homoduplex) A DNA molecule with completely complementary base sequences.

Homologies (French: homologies) Similarities in DNA or protein sequences between individuals of the same species or among different species.

Homologous chromosomes (French: chromosomes homologues) A pair of chromosomes containing the same linear gene sequences, each derived from one parent.

Homologous recombination, see: recombination homologous.

Homoplasmy (French: homoplasmie) The presence of a single population of mt DNA in the cells of a single individual. This is normal.

Homozygous (French: homozygote) Designating a diploid nucleus, cell or organism that contains two identical alleles for any one gene.

Host cell (French: cellule hôte) Cell carrying foreign genetic material brought by a virus, a plasmid, a recombine DNA in vitro or an entire cell.

Housekeeping gene (French: gène domestique) Gene that regulates vital functions of all cell types.

HPLC, high pressure liquid chromatography (French: HPLC, chromatographie liquide à haute performance) Chromatographic technology used to separate and quantitate mixtures of substances in solution. The high pressure technique allows a rapid analysis of complex mixtures.

Human gene therapy (French: thérapie génique humaine) Insertion of normal DNA directly into cells to correct a genetic defect.

Human Genome Initiative (French: initiative en génome humain) Collective name for several projects begun in 1986 by DOE to (1) create an ordered set of DNA segments from known chromosomal locations, (2) develop new computational methods for analyzing genetic map and DNA sequence data, and (3) develop new techniques and instruments for detecting and analyzing DNA. This DOE initiative supported by the NIH is now known as the Human Genome Program.

Hybrid DNA-RNA (French: hybride ADN-ARN) Double stranded molecule made of a DNA and a RNA complementary chains.

Hybrid gene, chimeric gene (French: gene hybride) Gene made of DNA fragments of various origins.

Hybrid selection (French: sélection d'hybride) Selection of a double-stranded DNA molecule with heteroduplex regions.

Hybridization (French: hybridation) The process of joining two complementary strands of DNA or one each of DNA and RNA to form a double-stranded molecule.

Hybridization, in situ (French: hybridation in situ) Hybridization of a specific DNA or RNA probe, marked with cellular DNA or RNA, on a tissue or fixed cells.

Hybridization on colony (French: hybridation sur colonie) Hybridization in situ allowing to identify bacteria with a specific DNA sequence.

Hydrocephaly (French: hydrocéphalie) Fluid invasion within the cranium. Abnormal enlargement of cerebral ventricles.

Hydrops, anasarca (French: hydrops) Fetus showing ascites and abnormal tissue oedema.

Hyperdiploidy (French: hyperdiploïdie) The state of being hyperploid, or having more than the typical number of chromosomes in unbalanced sets, as in Down's syndrome.

Hypertelorism (French: hypertélorisme) Abnormal distance between the two eyes measured at the pupil level.

Hypoploidy (French: hypoploïdie) State of having lost one or more chromosomes.

Hypothenar (French: hypothenar) The ridge on the palm along the basis of the fingers and the ulnar region.

I

Immunofluorescence technique, method (French : immunofluorescence, méthode) A method determining the location of antigen or antibody in tissue by the pattern of fluorescence resulting when the tissue is exposed to the specific antibody or antigen labelled with a fluorochrome.

Immunosuppression (French: immunosuppression) The artificial prevention or diminution of the immune response, as by irradiation or by administration of anti-metabolites, anti-lymphocyte serum, or specific antibody.

Imprinting, parental see: parental imprinting.

Inborn error of metabolism (French: erreur innée du métabolisme) Genetic metabolic disorder in which a protein defect produces a metabolic block which may have a deleterious effect.

Incidence (French: incidence) Number of new patients or individuals who acquired the disease during a certain period of time in a specific population.

Individual primary recombinant clones (French: recombinant individuel primaire) Recombinant clones (hosted in phage, cosmid, YAC or other vector that are placed in two-dimensional arrays in microtiter dishes. Each primary clone can be identified by the identity of the plate and the clone location (row and column) on that plate. Arrayed libraries of clones can be used for many applications, including screening for a specific gene or genomic region of interest as well as for physical mapping. Information gathered on individual clones from physical map analyses is entered into a relational database and used to construct physical and genetic linkage map simultaneously; clone identifiers serve to interrelate the multilevel maps.

Induction (French: induction) The starting or enhancement of synthesis of an enzyme by a cell, taking place upon the provision of the substrate for the enzyme.

Informatics (French: informatique) The study of the application of computer and statistical techniques to the management of information. In genome projects, informatics includes the development of methods to search databases quickly, to analyze DNA sequence information, and to predict protein sequence and structure from DNA sequence data.

Informativity (French: informativité) Possibility to identify two chromosomes of the same pair in particular in the region that carries the mutant gene.

Initiation factor (French: facteur d'initiation) Any class of protein factors which are requisite for the formation of the initiation complex and the initiation of mRNA translation.

Insert (French: insert) Sequence of foreign DNA introduced in a specific DNA molecule.

Insertion (French: insertion) The acquisition of extra nucleotides within a DNA sequence. They may affect only one nucleotide, a point mutation, but they usually affect several nucleotides in the sequence.

Insertion mutation, mutagenesis (French: mutagénèse par insertion) A mutation that results from

an insertion of one or more nucleotides into the DNA chain. They can revert to the wild-type state by removal of the extra sequences. They may change the triplet code reading of genetic information.

Insertion sequence, see : sequence, insertion.

In situ hybridization (French: hybridation in situ) Use of a DNA or RNA probe to detect the presence of the complementary DNA sequence in cloned bacterial or cultured eukaryote cells.

Integration (French: intégration) Recombination process that inserts a small DNA molecule into a larger one.

Intercalary (French: intercalaire) Loss of a portion of a chromosome between two breaks.

Interest gene (French: gène d'intérêt) Gene made of DNA fragments of various origins.

Interferon (French: interferon) A protein that is synthesized by animal cells in response to viral infection and non specifically inhibits replication of the viruses. It is found in serum almost at the onset of the infection and long before the production of antibody.

Interphase (French: interphase) The period in the cell cycle when DNA is replicated in the nucleus; it is followed by mitosis.

Interrupted gene (French: gène discontinu) Gene made of several alternate series of exons and introns RNA introns are eliminated during splicing. Differential splicing of introns of an interrupted gene can give rise to several proteins; one then speaks of mosaic gene.

Intragenic recombination (French: recombinaison intragénique) The process by which progeny derive a combination of genes different from that of either parent. In higher organisms, this can occur by crossing over.

Introns, intervening sequence (French: intron) The DNA base sequences interrupting the protein-coding sequences of a gene; these sequences are transcribed into RNA but are cut out of the message before it is translated into protein.

Inversion (French: inversion) An abnormality in chromosome structure that results from a portion of the chromosome becoming detached, rotating through 180o and then becoming attached again.

In vitro (French: in vitro) Occurring outside a living organism.

In vivo (French: in vivo) Taking place in the living body.

Isochromosome -i (French: isochromosome, -i) Abnormal chromosome formed by the duplication of two short arms -ip- or long arms -iq- of equal length and with identical loci. The other arm is lost.

Isoelectric focusing (French: électrofocalisation) An analytical separation procedure similar to gel electrophoresis. It is used to separate proteins and other charged molecules on the basis of their isoelectric point.

IVF, in vitro fertilization (French: fécondation in vitro, FIV) Fertilization of the ovum by a sperm outside of the living organism.

IVM, maturation in vitro (French: IVM, maturation in vitro) see maturation in vitro.

K

Karyotype (French: caryotype) A photomicrograph of individual chromosomes arranged in a standard format showing the number, size, and shape of each chromosome type; used in low- resolution physical mapping to correlate gross chromosomal abnormalities with the characteristics of specific diseases.

Kilobase, kb (French: kilobase, kb) Unit of length for DNA fragments equal to 1000 nucleotides.

Kinetochore, see: centromere.

L

LDL, low density cholesterol (French: cholesterol de basse densité, LDL) Cholesterol transported by low density lipoproteins.

Leaky (French: partiellement fonctionnel) Said of a gene mutation which allows some residual level of gene expression.

Leptonema (French: leptonème) First stage of the first meiotic division.

Lethal factor (French: facteur létal) An abnormality of the genome that leads to death in utero, ex :chromosomal anomaly, mutation.

Library (French: banque) An unordered collection of DNA clones from a particular organism whose relationship to each other can be established by physical mapping.

Ligand (French: ligand) A molecule that can bind to a receptor and thereby induce a signal in the cell, example: a hormone.

Ligase, DNA (French: ligase, ADN) An enzyme that creates a phosphodiester bond between the 3' end of one DNA segment and the 5'.

Ligation (French: ligature) Covalent linkage of a DNA molecule extremities allowing to reunite by a bridge two DNA molecules in one single unit.

Linkage (French: liaison, groupe de liaison) The proximity of two or more markers, genes or RFLP markers on a chromosome; the closer together the markers are, the lower the probability that they will be separated during DNA repair or replication processes (binary fission in prokaryotes, mitosis or meiosis in eukaryotes), and hence the greater the probability that they will be inherited together.

Linkage desequilibrium (French: déséquilibre de liaison) Situation where two genes or alleles are more often or less associated in cis in a given population and

for which the frequency departs from expected value according to the Hardy Weinberg law.

Linkage group (French: groupe de liaison) Group of genes that seem to be close together according to their hereditary transmission. They are generally neighbour on the same chromosome.

Linkage map (French: carte de liaison) A map of the relative positions of genetic loci on a chromosome, determined on the basis of how often the loci are inherited together. Distance is measured in centimorgans (cM).

Linker (French: lieur) Synthetic bicatenary olinucleotide added in vitro to a DNA sequence that brings a new restriction site.

Localize (French: localiser) Determination of the location of a gene or other marker on a chromosome.

Localized mutagenesis, see mutagenesis.

Locus (French: locus) The position on a chromosome of a gene or other chromosome marker; also means the DNA at that position. The use of locus is sometimes restricted to mean regions of DNA that are expressed.

Lod score (French: lod score) The abbreviation for logarithm of the odds. A measure of the odds ratio obtained by dividing the likelihood that two loci are linked at a specific recombination fraction by the likelihood that they are unlinked.

Lymphedema (French: lymphoédème) Congenital or acquired oedema due to an obstruction of lymphatic vessels.

Lymphocyte (French: lymphocyte) Leukocyte which arises in the lymph glands and the lymph nodes. It has a single nucleus and non-granular protoplasm.

Lyon hypothesis, see sex chromatin.

Lytic phage (French: phage de lyse) Any phage that causes host cells to lyse.

M

Macrocephaly (French: macrocéphalie) Excessive size of the head.

Macrorestriction map (French: carte de macrorestriction) Map depicting the order of, and distance between sites at which restriction enzymes cleave chromosomes.

Malformation (French: malformation) Any morphological defect present at birth, being apparent or not.

Mapping (French: cartographier) See gene mapping, linkage map, physical map.

Marker (French: marqueur) An identifiable physical location on a chromosome : restriction enzyme cutting site, gene whose inheritance can be monitored. Markers can be expressed regions of DNA (genes) or some segment of DNA with no known coding function but whose pattern of inheritance can be determined. Also see RFLP, restriction fragment length polymorphism.

Marker gene (French: gène marqueur) Gene expression that allows sorting of cells where it is found.

Maternal inheritance, see: cytoplasmic inheritance.

Maturase (French: maturase) Enzyme that intervenes in the maturation of pre-m-RNA.

Maturation (French: maturation) Modifications of messengerRNA made of additions and deletions at the time of, or after the transcription.

Maturation in vitro, IVM (French: maturation in vitro) To describe the maturation of ova in vitro.

Mb, see: megabase.

Megabase, Mb (French: mégabase, Mb) Unit of length for DNA fragments equal to 1 million nucleotides and roughly equal to 1 cM.

Meiosis (French: méiose) The process of two consecutive cell divisions in the diploid progenitors of sex cells. Meiosis results in four rather than two daughter cells, each with a haploid set of chromosomes.

Mendelian inheritance (French: hérédité mendélienne) Classical heredity as described by Gregory Mendel in 1866.

Mesoderm (French: mésoderme) One of the three layers of cells comprising the early embryo. Gives rise to muscle, bone, connective tissues and blood as well as the vascular and the uro-genital systems.

(French : ARN messager, ARNm) RNA that serves as a template for protein synthesis. Also see: genetic code.

Metacentric (French: métacentrique) Chromosome with a more or less central centromere and arms of approximately equal length.

Metaphase (French: métaphase) A stage in mitosis or meiosis during which the chromosomes are aligned along the equatorial plane of the cell.

Methylation (French: méthylation) Chemical reaction adding a methyl group to a compound. Note the hypermethylation in the FRA X syndrome leading to FMR1 gene inactivation. May be involved in the regulation of gene expression.

Microsatellite, minisatellite (French: microsatellite) Highly polymorphic DNA marker comprised of mononucleotides, dinucleotides, trinucleotides or tetra-nucleotides that are repeated in tandem arrays and distributed throughout the genome. The best studied are the CA <alternatively GT> dinucleotide repeats. They are used for genetic mapping.

Microcephaly (French: microcéphalie) Head circumference < 2nd percentile at birth. The other parameters can be normal.

Micrognathia (French: micrognathie) Congenital or acquired insufficient development of the mandible.

Midface (French: étage moyen de la face) Median region of the face.

Minicell (French: minicellule) Bacterial cell of reduced sized that has lost its chromosomal DNA.

Minichromosome (French: minichromosome) Unidentified small chromosome that replicates according to the mitotic cell cycle. Generally there is only one copy of this chromosome.

Minigene (French: minigène) Gene, reconstructed for experimental means, from regulating sequences and double strand cDNA and the corresponding mRNA. A minigene codes directly for mature mRNA.

Miniphage (French: miniphage) Phage, reconstructed for experimental needs, that has kept part of its functions.

Miniplasmid (French: miniplasmide) Plasmid reconstructed for experimental needs with remaining part of its functions.

Misense mutation (French: mutation non-sens) mutation that replaces an amino acid codon by a nonsense codon.

Mismatch, misparing (French: mésappariement) Non pairing of a zone in a fragment of nucleic acid double strand.

Mitogen (French: mitogène) Agent inducing mitoses.

Mitosis (French: mitose, caryocinèse) The process of nuclear division in cells that produces daughter cells genetically identical to each other and to the parent cell.

Mitotic index (French: index mitotique) Proportion of dividing cells in relation to all cells analyzed.

Modification, nucleic acid (French: modification d'un acide nucléique) Any transformation observed in nucleotides after their assembling into a polynucleotide.

Mixoploidy, see: mosaicism.

Modal number (French: nombre modal) Normal number of chromosomes.

Molecular biology (French: biologie moléculaire) Study of molecules carrying the hereditary message, DNA,RNA structure, synthesis, modifications or transformations.

Molecular genetics (French: génétique moléculaire) Branch of genetics concerned with the molecular structure and activities of the genetic material, including the replication of DNA, the transcription into RNA and the translation of RNA to form proteins.

Molecular hybridization, see : hybridization.

Monocistronic (French: monocistronique) Descriptive of messenger RNA molecules that code for one polypeptide chain.

Monoclonal antibodies (French: anticorps monoclonaux) Homogeneous antibodies produced by a clone of B lymphocytes originating from a unique mother cell that will generally detect only one genetic determinant.

Monolayer (French: monocouche) Tissue culture in monocellular layers.

Monosomy (French: monosomie) Presence of only one copy of a pair or segment of a chromosome.

Monozygocity (French: monozygotie) Twins originating from the same egg.

Morgan, see: centimorgan.

Morphogenesis (French: morphonogénèse) Development of forms and structures of the organism.

Morula (French: morule) Name given to the human fertilized egg after the first divisions or blastomere segmentation. At this stage the egg is still in the genital tract. Nidation takes place at the blastocyst stage.

Mosaic (French: mosaïque, myxoploïdie) Presence of two or more distinct cells in an individual.

mRNA, see messenger RNA.

Mucolipidosis (French: mucopolidose) Group of hereditry metabolic defects due to the deficiency of enzymes essential for the degradation of oligosaccharides; unlike mucopolysaccharidoses they do not excrete abnormal amounts of glysaminoglycans in urine. Skeletal, cardiac, ocular and systemic deleterious effects are observed in the affected individuals.

Multifactorial or multigenic disorders (French: maladies multifactorielles, multigéniques) See polygenic disorders.

Multigeny (French: multigénie) Genetic property that presents a phenotype character that is dependant of several genes.

Multiple allelomorphs (French: alleles multiples) Relative to various possible forms of a gene.

Multiplexing (French: multiplexe) A sequencing approach that uses several pooled samples simultaneously, greatly increasing sequencing speed.

Mutagen (French: mutagène) Any physical or chemical agent significantly increasing mutational events and thus mutation rates above the spontaneous background level. Those are induced mutations as opposed to the normally occurring spontaneous mutations.

Mutagenic (French: mutagène) Capable of inducing stable, heritable changes in the genetic information of a cell.

Mutagenic agent, see mutagen.

Mutant (French: mutant) Gene that has undergone a mutation or an individual who carries this mutant gene.

Mutation (French: mutation) Any heritable change in DNA sequence. Compare polymorphism.

N

NAIP, see: neuronal apoptosis inhibitory protein.

Nearest neighbor sequence analysis (French: A biochemical technique for estimating the frequencies that pairs are next to one another.

Neuronal apoptosis inhibitory protein, NAIP (French: Protéine inhibitrice de la mort neuronale programmée) Protein that inhibits the programmed neuronal death.

Neurotropic factors (French: facteurs neurotropiques) Factors that have some affinity for the nervous tissues.

Nick (French: cassure monocaténaire) Breakage affecting only one of two strands of a nucleotide double helix.

Nick translation (French: marquage par déplacement de l'encoche) Replaces part of a preexisting strand of duplex DNA with newly synthesized material.

Nitrogen base (French: base nitrogénique) A nitrogen-containing molecule having the chemical properties of a base.

Non disjunction (French: non disjonction) Failure of separation of homologue chromatids at the mitotic anaphase as well as the second meiotic division. There is desequilibrium of the two sister alleles or chromosomes.

Nonsense codon (French: codon nonsens) Codon that does not specify an amino acid but indicates the termination of a polypeptide chain. These codons interrupt the reading of the messenger RNA (mRNA) strand and also cause release of the synthesised polypeptide chain.

Nonsense triplet, see: stop codon.

Northern blot, RNA transfer (French: Northern blot, transfert d'ARN) A gel base procedure that locates mRNA sequences on a gel that are complementary to a piece of DNA used as a probe.

Nucleic acid (French: acide nucléique) A large molecule composed of nucleotide subunits.

Nucleic acid denaturation (French: dénaturation d'acide nucléique) Conversion of double stranded nucleic acid to a single strand state.

Nucleic acid sequencing (French: décryptage des séquences nucléotidiques) Determination of the linear order of the nucleic basic chains.

Nucleic probe (French: sonde nucléique) DNA or RNA sequence marked by a fluorescent isotope or enzyme used to detect homologue sequences by in situ or in vitro hybridization.

Nucleosome (French: nucléosome) Element of chromatin made by the coiling of 140 base pairs on an octamere of histone.

Nucleotide (French: nucleotide) A subunit of DNA or RNA consisting of a nitrogenous base (adenine, guanine, thymine, or cytosine in DNA; adenine, guanine, uracil, or cytosine in RNA), a phosphate molecule, and a sugar molecule (deoxyribose in DNA and ribose in RNA). Thousands of nucleotides are linked to form a DNA or RNA molecule. See DNA, base pair, RNA.

Nucleus (French: noyau) The cellular organelle in eukaryotes that contains the genetic material.

Null mutation (French: mutation nulle) An allele that results in either the absence of the gene product or the absence of any function at the phenotypic level.

O

Oligogenic diseases (French: maladies oligogéniques) Diseases or traits that result from the effects of relatively few genes some of which have relatively large effects.

Omphalocele (French: omphalocèle) Umbilical hernia.

Oncogene (French: oncogène) A gene, one or more forms of which is associated with cancer. Many oncogenes are involved, directly or indirectly, in controlling the rate of cell growth.

Oocyte (French: ovocyte, oocyte) An immature or unfertilized ovum, also called the egg.

Open reading frame (French: cadre de lecture ouvert, ORF) The sequence of DNA or RNA located between the start-code sequence, or initiation codon and the stop-code sequence, or termination codon.

Operator (French: opérateur) DNA site to which a repressor will bind to prevent the transcription of the adjacent promotor.

Operon (French: opéron) Transcription unit made of a promotor, operator and one or more structural genes.

ORF, see: open reading frame.

Overlapping clones (French: clones superposés) See genomic library.

Overlapping sequence, see: sequence overlapping.

Ovum (French: ovum) A female germ cell; an egg cell; a cell which is capable of developing into a new member of the same species, in animals usually only after maturation and fertilization.

P

Pachytene (French: pachytène) Genetic viral material inside an infected cell Qualifies The third stage of the first meiotic division when chromosomes are condensed , chromatids of each chromosome are visible. Homologue chromosomes appear as bivalent.

Packaging (French: encapsidation) Packaging of genetic viral material inside an infected cell.

Palindromic sequence (French: sequence palindromique) A nucleotide sequence in which the 5' to 3' sequence of one strand is the same as that of its complementary strand.

Panmixia (French: panmixie) Large human population where unions between individuals take place at random. Genotypes depend on the gene and allele frequencies.

Parental coefficient (French: coefficient de parenté) The probability that an individual has received both alleles of a pair from an identical ancestral source. Also the proportion of loci at which an individual is homozygous by descent.

Parental imprinting (French: empreinte parentale) The differential expression of genetic material, at either a chromosomal or an allelic level, depending on whether the genetic material has been inherited from the male or female patient.

Patent ductus arteriosus (French: persistance du canal artériel) Congenital heart defect : persistance of the arterial canal.

Patent foramen ovale (French: communication inter-auriculaire) Congenital heart defect: persistance of the inter auricular communication.

PCR, see polymerase chain reaction.

PDG, preimplantation diagnosis (French: diagnostic prenatal pré-implantatoire) Diagnosis of a disease before the egg implantation.

Pedigree, see family tree.

Penetrance (French: pénétrance) Frequence with which a trait will manifest in a family.

Pentasomy (French: pentasomie) State of a cell or individual that has five copies of a chromosome instead of two. Example: the 49 XXXXX syndrome.

PEPCK, phosphoenolpyruvate carboxylase enzyme (French: enzyme phosphoenolpyruvate carboxylase)

Peptide signal, Peptide sequence (French: peptide, séquence) Segment of 15 to 30 amino acids present at the N terminal portion of a protein; it indicates to the cell that this protein must be exported or secreted.

PERV virus (French: virus PERV) Pork virus known as the endogen pork virus found in the pork genetic material. This retrovirus can not be identified or eliminated and the laboratory tests showed that it can infect human cells. We do not know if the retrovirus can be transmitted via a xenograft.

Phage (French: phage) A virus for which the natural host is a bacterial cell.

Phage template (French: phage tempéré) Phage genome can be integrated into the host cell DNA and transform its properties. 1- if integrated into the host cell genome it is called prophage and 2- the template phage has the capacity to lyse the bacteria that it infects.

S phase (French: S phase) Interphase which usually lasts much longer than mitosis is itself divided into stages. Of the interphasic stages, Gap 1 (G1) which lies between the end of telophase and the beginning of the Synthesis (S) periods is the longest; DNA synthesis takes place during the S period.

Phenocopy (French: phénocopie) A mimic of a phenotype that is usually determined by a specific genotype, produced instead by the interaction of some environmental factor with a different genotype.

Phenotype (French: phénotype) The biochemical physiological and morphological characteristics of an individual as determined by his or her genotype and the environment in which it is expressed.

Philtrum (French: philtrum) Region of the upper lip bordered by two tissue pillars.

Phosphodiester (French: phosphodiester) Molecule that has one phosphate and two ester functions.

Physical map (French: carte physique) A map of the locations of identifiable landmarks on DNA (e.g., restriction enzyme cutting sites, genes), regardless of inheritance. Distance is measured in base pairs. For the human genome, the lowest- resolution physical map is the banding patterns on the 24 different chromosomes including X and Y chromosomes the highest-resolution map would be the complete nucleotide sequence of the chromosome.

Phytohemagglutinin (French: phytohémagglutinine) Polysaccharide substance extracted from red beans that has the capacity to agglutinate red cells and facilitate their separation from leukocytes. It also has the property to stimulate lymphocyte division and blastic transformation. This property is used in cytogenetic studies done on cultured lymphocytes.

Pierre Robin syndrome (French: syndrome de Pierre Robin) Syndrome involving the abnormal closure of the palate and the inferior mandible that is too small.

Placebo (French: placebo) Inactive substance substituted for a medication in order to distinguish between the psychological and pharmacological effects.

Placental mosaicism, see: confined placental mosaicism.

Plasmid (French: plasmide) Autonomously replicating, extra chromosomal circular DNA molecules, distinct from the normal bacterial genome and non essential for cell survival under non selective conditions. Some plasmids are capable of integrating into the host genome. A number of artificially constructed plasmids are used as cloning vector.

Plasmid incompatibility (French: incompatibilité plasmidique) The inability of two different plasmids to coexist in the same host cell.

Plasmid Ri (French: plasmide Ri) Plasmid carried by the bacteria Agrobacterium rhizo-genes that has a transferable DNA segment and that can be integrated into the genome of a vegetable host cell. It can serve as transformation vector to produce transgenic plants.

Plasmide Ti (French: plasmide Ti) Plasmid carried by the bacteria Agrobacterium rhizo-genes that has a DNA segment transfeerable and has the property to

integrated into the DNA of a vegetable host cell . Ti means tumor inductor.

Pleiotropy (French: pléïotropie) Multiple phenotypic effects of a single gene or gene pair. The term is used particularly when the effects are ordinarily thought to be unrelated.

Ploidy (French: ploïdie) A term referring to the number of chromosome sets per cell. Example: haploid; diploid; polyploid.

Pluripotent stem cell, see: stem cell.

Point mutation (French: mutation ponctuelle) Mutation interesting only one base. Addition or deletion are mutation mechanisms.

Polar body (French: globule polaire) Hypoploid cell resulting from the first two meiotic divisions. It contains mainly nuclear material, the cytoplasmic material being reserved for the ovocyte and the ovotid.

Polar mutation (French: mutation polaire) Mutation in one gene that reduces the expression of one gene further from the promoter in the same operon. Nonsence mutations frequently are polar.

Poly A tail, polyadenylated end, polyA region (French: séquence polyA) Long segment of adenosine polymerized monophosphates present at the 3' end of eukaryotes mRNAs.

Polydactyly (French: polydactylie) Presence of one or more fingers, or duplication of one finger or toe.

Polygenic disorders (French: maladies polygéniques) Genetic disorders resulting from the combined action of alleles of more than one gene like heart disease, diabetes, and some cancers). Although such disorders are inherited, they depend on the simultaneous presence of several alleles; thus the hereditary patterns are usually more complex than those of single- gene disorders.

Polymerase chain reaction, PCR (French: réaction en chaîne de la polymérase, PCR) A method for amplifying a DNA base sequence using a heat-stable polymerase and two 20- base primers, one complementary to the (+)- strand at one end of the sequence to be amplified and the other complementary to the (-)- strand at the other end. Because the newly synthesized DNA strands can subsequently serve as additional templates for the same primer sequences, successive rounds of primer annealing, strand elongation, and dissociation produce rapid and highly specific amplification of the desired sequence. PCR also can be used to detect the existence of the defined sequence in a DNA sample.

Polymerase, DNA or RNA (French: polymérase, ADN ou ARN) Enzymes that catalyze the synthesis of nucleic acids on preexisting nucleic acid templates, assembling RNA from ribonucleotides or DNA from deoxyribonucleotides.

Polymorphism (French: polymorphisme) Difference in DNA sequence among individuals. Genetic variations occurring in more than 1% of a population would be considered useful polymorphisms for genetic linkage analysis.

Polypeptide chain termination (French: Terminaison de la chaîne peptidique) End of the proteic chain.

Polyploidy (French: polyploïdie) Cells, tissues or individuals that have more than a diploid set of chromosomes. Triploid and tetraploid embryos are non viable, they are often found in spontaneous abortions.

Polyribosome (French: polyribosome) Complex made of a mRNA molecule and ribosomes. The protein synthesis takes place on this complex.

Prader Willi syndrome, see: Angelman syndrome.

Precursor RNA, see pre-messenger RNA.

Preimplantation diagnosis, see PDG.

Pre messenger RNA, Heterogeneous nuclear RNA (French: Pré ARN messager) The probable precursor of messenger RNA, mRNA, so named because its size distribution is heterogeneous and its location is strictly nuclear; no base sequences have been found in cytoplasmic mRNA that are not also pre-messenger RNA.

Premutation (French: prémutation) Stage of expansion mechanism in which the mucleotide expansion is above normal but still insufficient to translate into clinical manifestations. For instance in myotonic dystrophy a 50 to 80 triplet repeat is considered as a premutation.

Prevalence (French: prévalence) Frequency of a disease in a population, counting both old and new cases.

Primer (French: amorce) Short preexisting polynucleotide chain to which new deoxyribonucleotides can be added by DNA polymerase.

Proband, see propositus.

Probe (French: sonde) Single-stranded DNA or RNA molecules of specific base sequence, labeled either radioactively or immunologically, that are used to detect the complementary base sequence by hybridization.

Processing RNA, post transcriptional processing (French: maturation de l'ARN) Modifications of messengerRNA made of additions and deletions at the time of transcription.

Prognathism (French: prognathisme) Excessive development of the lower jaw.

Prokaryote (French: procaryote) Cell or organism lacking a membrane- bound, structurally discrete nucleus and other subcellular compartments. Bacteria are prokaryotes.

Promoter (French: promoteur) A site on DNA to which RNA polymerase will bind and initiate transcription.

Prophage (French: prophage) Sequence of phage DNA.

Propositus, proband (French: proband, propositus) The family member through whom the family is ascertained. If the propositus is affected, he may be called the index case.

Prosome (French: prosome) Small RNA particle associated with a free messenger RNA repressed in the cytoplasm.

Protein (French: protéine) A large molecule composed of one or more chains of amino acids in a specific order; the order is determined by the base sequence of nucleotides in the gene coding for the protein. Proteins are required for the structure, function, and regulation of the body cells, tissues, and organs, and each protein has unique. Examples are hormones, enzymes, and antibodies.

Protein design (French: remodelage) Creation of a protein with new properties by directed mutagenesis or gene synthesis.

Protein, structural (French: protéine de structure) A protein that serves a structural role in the body such as collagen.

Proteomics (French: protéomique) New discipline derived from genomics that concerns research activities aimed at the collection of all available information on gene expression of organisms with an identified genome.

Protooncogen (French: protooncogène) A normal gene that with a slight alteration by mutation or other mechanism becomes an oncogene.

Provirus (French: provirus) The state of a virus in which it integrates into and replicates in coordination with a host cell chromosome and thus is transmitted from one cell generation to another.

Pseudoautosomal (French: pseudoautosomique) The distal tip of the Y chromosome short arm, which undergoes crossover with the distal tip of the X chromosome short arm during meiosis in the male.

Pseudodiploidy (French: pseudodiploïdie) Apparent diploid cell. The karyotype is abnormal, although it carries 46 chromosomes.

Pseudogene (French: pseudogène) The sequence of pseudogene is similar to the structural gene but does not code for a protein.

Pseudohermaphrodite (French: pseudohermaphrodite) Individual whose gonadal and phenotypic sex differ.

Pterygium colli, webbed neck (French: pterygium colli, cou palmé) A thick fold of skin on the lateral side of the neck, extending from the mastoid region to the acromion, producing a congenital webbed neck.

Ptosis (French: ptose) Paralytic drooping of the upper eyelid.

Puff, see: chromosome puff.

Pulse labelling (French: marquage de brève durée) An experimental technique in which cells or cell extracts are exposed for a short time to a compound labelled with a radio active nucleotide.

Purine (French: purine) A nitrogen- containing, single-ring, basic compound that occurs in nucleic acids. The purines in DNA and RNA are adenine and guanine.

Purine base (French: base purique) Adenine and Guanine.

Pyrimidic base (French: base pyrimidique) Cytosine and Thymine.

Pyrimidine (French: pyrimidine) A nitrogen-containing, double- ring, basic compound that occurs in nucleic acids.The pyrimidines in DNA are cytosine and thymine; in RNA, cytosine and uracil.

Q

Quadriradial pattern (French: image quadriradiale) Structure shaped as a cross made of homologue or non homologue chromosomes observed when a translocation takes place during a mitotic division.

Quasidominance (French: quasidominance) The pattern of inheritance produced by the mating of an affected homozygote individual heterozygous for the same recessive trait so that homozygous affected members appear in two or more successive generations.

R

Rare-cutter enzyme, see restriction enzyme cutting site.

Receptor (French: récepteur) A transmembrane or intracellular protein involved in transmission of a cell signal.

Recessive (French: récessif) A trait or gene that is only expressed in homozygotes or hemizgotes.

Recombinant (French: recombiné) An individual who has a new recombination of genes not found together in either parent. Usually applied to linkage analysis.

Recombinant aneusomia (French: aneusomie de recombinaison) Presence of a duplication or deletion resulting from a crossing over in an inverted loop.

Recombinant clones (French: clones recombinants) Clones containing recombinant DNA molecules. See recombinant DNA technologies.

Recombinant DNA molecules (French: molécules d'ADN recombinant) A combination of DNA molecules of various origins that are joined using recombinant DNA technologies.

Recombinant DNA technologies (French: technologies d'ADN recombinant) Procedures used to join together DNA segments in a cell- free system (an environment outside a cell or organism). Under

appropriate conditions, a recombinant DNA molecule can enter a cell and replicate there, either autonomously or after it has become integrated into a cellular chromosome.

Recombinant genetic (French: recombinant génétique) Any of the individual or cells arising as a result of inter-chromosomal and intra-chromosomal,via crossing-over or conversion, genetic recombination.

Recombinant plasmid (French: plasmide recombiné) Plasmid in which a foreign DNA fragment has been inserted.

Recombinant protein (French: protéine recombinée, recombinante) Protein synthesised from a transgene.

Recombination frequencies (French: fréquences de recombinaison) Instance when the progeny derive a combination of genes different from that of either parent. In higher organisms, this can occur by crossing over.

Recombination, homologous (French: recombinaison homologue) Insertion of a transgene in a particular site in lieu of a of a nervous cell specific gene.

Recombination, intragenic (French: recombinaison intragénique) The process by which progeny derive a combination of genes different from that of either parent. In higher organisms, this can occur by crossing over.

Regulation, autogenous (French: régulation autogène) Regulation system where the gene product controls its own expression.

Regulation mutation (French: mutation de régulation) Mutation that affects the regulation of one or more gene expression, without affecting the coding segment.

Regulatory gene (French: gène de régulation) A gene coding for a protein that regulates other genes.

Regulatory regions or sequences (French: régions régulatrices ou séquences) A DNA base sequence that controls gene expression.

Reject (French: rejet) Immunological reaction against a tissue or organ. Elimination by the receiver of the transplanted tissue or organ.

Release factor (French: facteur de terminaison) Any of the special protein factors which recognize the terminator codons UAA, UAG, and UGA in messenger RNA and stimulate the codon specific release of polypeptides from ribosomes during genetic translation.

Renaturation of nucleic acid (French: renaturation d'acide nucléique) The return by slow conversion of a denatured nucleic acid or protein to its native configuration.

Repeat unit (French: unité de répétition)DNA sequence constituting the basic motive in a repeated region.

Replication fork (French: fourche de réplication) A Y shaped point at which two strands of a DNA molecule are unwound and separated during replication.

Replicon (French: réplicon) Any genetic element - bacterial chromosome, virus, genome, plasmid - that behaves as an autonomous unit of replication.

Repressor (French: répresseur) Protein synthesised by a regulator gene, which, by binding to a specific site on DNA, the operator gene of an operon, prevents the formation of messenger RNA by the operon's other structural genes and hence stops protein -enzyme-synthesis.

Reproduction, assisted technology ART (French: reproduction assistée, ART) (assisted

reproduction technology, ART) Technology that concerns one or several steps of intervention during the process of in vitro reproduction.

Resistance factor, R factor, R plasmid, resistance plasmid (French: facteur de transfert de résistance) Plasmid that codes for one or more enzymes inactivating one or more toxic agents or antibiotics.

Resolution (French: résolution) Degree of molecular detail on a physical map of DNA, ranging low to high.

Restriction (French: restriction) Mechanism by which a cell degrades foreign DNA.

Restriction enzyme cutting site (French: site de coupure de l'enzyme de restriction) A specific nucleotide sequence of DNA at which a particular restriction enzyme cuts the DNA. Some sites occur frequently in DNA (e.g., every several hundred base pairs), others much less frequently (rare- cutter; e.g., every 10,000 base pairs).

Restriction enzyme, endonuclease (French: enzyme de restriction, endonucléase) A protein that recognizes specific, short nucleotide sequences and cuts DNA at those sites. Bacteria contain over 400 such enzymes that recognize and cut over 100 different DNA sequences.

Restriction fragment length polymorphism, RFLP (French: Variation between individuals in DNA fragment sizes cut by specific restriction enzymes; polymorphic sequences that result in RFLPs are used as markers on both physical maps and genetic linkage maps. RFLPs are usually caused by mutation at a cutting site.

Retinoblastoma (French: rétinoblastome) The retinoblastoma, uni or bilateral is an embryonic tumor of the retina. The gene locus for the retinoblastoma is located in the 13q14.1-14.2 region. In the proximal region of the long arm of chromosome 13. The tumor can be hereditary due to a germinal mutation or sporadic due to a somatic mutation.

Retrognathism (French: rétrognathisme) Small inferior mandible in a retro position.

Retrograde transport (French: transport rétrograde) Transport going in the other direction. Example: from the muscle to the nerve.

Retroinhibition (French: inhibition par rétroaction) Regulator system in which the terminal product of a biosynthetic chain of reactions stops the activity of the first enzyme of this chain.

Retrotransposon (French: rétrotransposon) Class of transposon of which the transposition requires the inverse transcription of their transcription product

Retrovirus (French: rétrovirus) A virus with an RNA genome that propagates by conversion of the RNA into DNA by the enzyme reverse transcriptase.

Reverse genetics (French: génétique réverse) The application of human gene mapping to clone the gene responsible for a particular disease when no information about the biochemical basis of the disease is available.

Reverse mutant (French: mutant réverse) Organism resulting from a reverse mutant.

Reverse transcriptase (French: réverse transcriptase) An enzyme used by retroviruses to form a complementary DNA sequence, cDNA, from their RNA. The resulting DNA is then inserted into the chromosome of the host cell.

Reversion (French: réversion) Mutation that restores a function annulled by a first mutation.

RFLP, see: restriction fragment length polymorphism.

RI plasmid, see plasmid RI.

Ribonucleic acid, RNA (French: acide ribonucléique, ARN) A chemical found in the nucleus and cytoplasm of cells; it plays an important role in protein synthesis and other chemical activities of the cell. The structure of RNA is similar to that of DNA. There are several classes of RNA molecules, including messenger RNA, transfer RNA, ribosomal RNA, and other small RNAs, each serving a different purpose.

Ribonucleotides, see nucleotide.

Ribosomal proteins (French: proteins ribosomales) A group of protein slinked to rRNA by non covalent bomds giving to the ribosome a tridimention structure.

Ribosomal RNA, rRNA (French: ARN ribosomal, ARNr) A class of RNA found in the ribosomes of cells.

Ribosomes (French: ribosomes) Small cellular components composed of specialized ribosomal RNA and protein; site of protein synthesis. See ribonucleic acid (RNA).

Ring chromosome (French: chromosome en anneau) Structurally abnormal chromosome in which the end of each chromosome arm has been broken and the broken arms reunited to form a ring.

RNA, see ribonucleic acid.

RNA processing, see: processing.

RNA replicase (French: ARN synthétase) ARN réplicase (RNA replicase) Enzyme of RNA viruses that catalyses the reproduction or replication of this molecule.

RNA satellite (French: ARN satellite) RNA that may be found in some viruses.

S

Sanger method, Sanger sequencing (French: méthode de Sanger) A widely used method to determine the order of bases in DNA.

Satellite DNA (French: satellite, ADN) Molecular: a portion of DNA that differs enough in base composition so that it forms a distinct band on cesium chloride gradient centrifugation; usually contains highly repetitive sequences. Cytogenetics: small mass of chromatin located at the extremity of the short arm of acrocentric chromosomes above a small constriction.

Segregation (French: ségrégation) Separation of two alleles on a locus at meiosis.

Sequence (French: séquence) See base sequence.

Sequence amplified (French: séquence amplifiée) Increased number of intra or extrachromosomic DNA sequences.

Sequence, coding (French: séquence codante) Segment of a gene that directly defines the amino acid sequence of the corresponding protein.

Sequence consensus (French: séquence consensus) Specific sequence of a given region of a nucleic acid or a protein in which each position represents the base or the amino acid most commonly found.

Sequence highly repeated (French: séquence hautement répétée) DNA sequence present in a great number of copies in the genome.

Sequence, insertion (French: séquence d'insertion) DNA element showing transposition from one region of the genome to another.

Sequences, inverted repeat (French: séquences répétées inverses) Identical or almost identical sequences present in several copies in the same DNA molecule with an inverse orientation.

Sequence leader (French: séquence de tête) Sequence found upstream of the initiation codon of traduction of RNA messengers.

Sequence, non coding (French: séquence non codante) Part of a gene that does not directly define the amino acid sequence of the corresponding protein.

Sequence, overlapping (French: séquence chevauchante) DNA sequence carrying the information related to several genes using a different reading frame.

Sequence, palindromic (French: séquence palindromique) Sequence of DNA that is the same when one strand is read right to left; consists os adjacent inverted repeats.

Sequence poly A (French: séquence polyA) Long segment of polymerized adenosine monophosphates present at the 3'extremity of mRNA of eukaryotes.

Sequence repeat (French: séquences répétées directes) Multiple copies of the same base sequence on a chromosome; used as a marker in physical mapping.

Sequence signal (French: séquence signal) DNA sequence that can be read the same way in both directions in relation to a central point : on the same strand example : ATTGC,CGTTA, or on both strands AAGTT and TTGCAA.

Sequence tagged site, STS (French: courte séquence d'ADN) Short (200 to 500 base pairs) DNA sequence that has a single occurrence in the human genome and whose location and base sequence are known. Detectable by polymerase chain reaction, STSs are useful for localizing and orienting the mapping and sequence data reported from many different laboratories and serve as landmarks on the developing physical map of the human genome. Expressed sequence tags (ESTs) are STSs derived from cDNAs.

Sequence, tandem repeat (French: séquences répétées en tandem) Multiple copies of the same base sequence on a chromosome; used as markers in physical mapping.

Sequence, unique (French: unique séquence) Selection of a monocatenary nucleic acid after formation of a hybrid molecule with a complementary strand.

Sequence untranslated (French: séquence non traduite) A region of mRNA that is not used in the synthesis of an amino acids sequence of a given peptide or protein. They usually appear at either end of the sequence that codes for the amino acid sequence.

Sequencing (French: séquençage) Determination of the order of nucleotides (base sequences) in a DNA or RNA molecule or the order of amino acids in a protein.

Sex chromatin, Barr body (French: chromatine sexuelle, corpuscule de Barr) Chromatin mass present in interphase nuclei of women and most female mammals representing one of the two Xs that is inactivated.

Sex chromosomes (French: chromosomes sexuels) The X and Y chromosomes in human beings that determine the sex of an individual. Females have two X chromosomes in diploid cells; males have an X and a Y chromosome. The sex chromosomes comprise the 23rd chromosome pair in a karyotype.

Shotgun cloning or method (French: méthode à l'aveugle) Cloning of DNA fragments randomly generated from a genome Sequencing method that involves randomly sequenced cloned pieces of the genome, without knowledge of where the piece originally came from. This can be contrasted with "directed" strategies, in which pieces of DNA from known chromosomal locations are sequenced. Because there are advantages to both strategies, researchers use both random (or shotgun) and directed strategies in combination to sequence the human genome.

Siblings (French: germains) Brothers and sisters; couple's own children.

Sibship (French: fratrie) All the sibs in a family.

Signal peptide, see peptide sequence.

(French: silenceur) DNA region located near a gene that has the capacity to reduce its transcription.

Silent mutation (French: mutation silencieuse) A mutant gene that has no phenotypic effect.

Simian crease (French: pli simiesque) Transverse and unique palmar crease frequently found in trisomy 21.

Single-gene disorder (French: maladie monogénique) Hereditary disorder caused by a mutant allele of a single gene. Examples: Duchenne muscular dystrophy, retinoblastoma, sickle cell disease.

Single stranded loop (French: boucle monocaténaire à brin unique) Non pairing loop.

Sister chromatid exchange, SCE (French: échange de chromatides soeurs) Exchange of equivalent material between two chromatids of a maternal or paternal chromosome.This is not an exchange between homologue chromosomes and it happens only in the first meiotic division by crossing over.

Site specific mutagenesis (French: mutagénèse dirigée) Introduction of a precise mutation in a cloned DNA fragment followed by the reinsertion of the mutant sequence in the original gene to replace the corresponding wild DNA.

SKY, see: Spectral karyotype SKY.

SMA, spinal muscular atrophy (French: amyotrophie spinale infantile) Neuromuscular disease.

SMN Survival motor neurone.

SNRNP, small nuclear ribonucleoprotein (French: petite ribonucléoprotéine nucléaire) Protein induced in the cytoplasm, that has affinity with the survival motor neuron, that plays a role in DNA splicing.

Somatic cell genetic mutation A change in the genetic structure that is neither inherited nor passed to offspring. Also called acquired mutations.

Somatic cell mutation (French: mutation somatique) A change in the genetic structure that is neither inherited nor passed to offspring. Also called acquired mutation.

Somatic cells (French: cellules somatiques) Any cell in the body except gametes and their precursors.

Somatic mutation (French: mutation somatique) Mutation in a cell that is not germinal.

Southern blotting (French: marquage Southern) Transfer by absorption of DNA fragments, separated in

electrophoretic gels, to membrane filters for the detection of specific base sequences by radio labelled complementary probes.

Spacer, DNA (French: espaceur) Untranslated DNA sequence separating genes in repeat units.

Species (French: espèce) Branch of natural sciences that concerns similar individuals that can reproduce within themselves.

Spectral karyotype SKY A graphic of all an organism's chromosomes, each labelled with a different color. Useful for identifying chromosomal abnormalities.

Sperm (French: sperme) The male sex cell

Spinal (French: spinal) Pertaining to the spine or vertebral column

Spliceosome (French: spliceosome) Ribonucleoproteic protein formed at the time of transcripts splicing

Splicing (French: épissage) The introduction of donor DNA into a vector for cloning

Splicing, mRNA (French: épissage, ARNm) The natural process by which transcribed mRNA matures to become mRNA that will be translated.

Staining region homogeneously (French: région de coloration homogène) Chromosome region that has a uniform staining property.

Initiation codon (French: codon d'initiation) A codon that codes for the first amino acid in a polypetide sequence.

Stem cell (French: cellule souche) Pluripotent cell giving rise to cells with a different function. The origin can also be embryonic or germinal. Example: undifferentiated, primitive cells in the bone marrow that have the ability both to multiply and to differentiate into specific blood cells.

Stenosis, arterial (French: sténose artérielle) Narrowing of an arterial vessel.

Stop codon (French: codon non-sens) see nonsense codon.

Stop signal (French: signal stop) Signal indicating the end of a gene.

Structural gene (French: gene de structure) A gene coding for any RNA or protein product.

Structural genomics (French: génomique de structure) The effort to determine the 3D structures of large numbers of proteins using both experimental techniques and computer simulation.

STS, see sequence tagged site.

Submetacentric (French: submétacentrique) A chromosome with an off-center centromere and arms of clearly different lengths.

Substitution (French: substitution) In genetics, a type of mutation due to replacement of one nucleotide in a DNA sequence by another nucleotide or replacement of one amino acid in a protein by another amino acid.

Suppression intergenic, mutation (French: suppression intergénique) Recuperation of a lost function with the help of a second mutation localized on a gene other than the initial mutant.

Suppression intragenic (French: suppression intragénique) A compensation mutation inside the mutant gene that will that will restaure its activity. The second mutation interests the first gene mutation but in a different site.

Suppressor gene A gene that can suppress the action of another gene.

Suppressor mutation (French: mutation suppressive) Gene action suppressed by another gene.

Synapsis (French: appariement, synapse) Pairing between homologous chromosomes of maternal and paternal origin during the prophase of meiosis, leading to the formation of gametes. Contact zone between two neurones or one neurone and a muscular glandular cell, through which the electric or chemical transmission is completed.

Synaptonemal complex (French: complexe synaptonémal) Electron micrographs of paired chromosomes at the pachytene stage of meiosis.

Syndrome (French: syndrome) The group or recognizable pattern of symptoms or abnormalities that indicate a particular trait or disease.

Syngeneic (French: syngénique) Genetically identical members of the same species.

Synteny (French: synténie) The physical presence together on the same chromosome of two or more gene loci, whether or not they are close enough together for linkage to be demonstrated.

System, acellular (French: système acellulaire) void of cells but containing the necessary elements for a specific synthesis, like nucleic acids, precursors, enzymes except substances to be tested in tissue extracts.

T

Tandem repeat sequences (French: séquences répétées en tandem) Multiple copies of the same base sequence on a chromosome; used as a marker in physical mapping.

TATA box (French: boîte TATA) A conserved A-T rich heptamer found about 25 bp before the start point of each eucaryotic RNA polymerase II transcription unit.

Technology transfer (French: transfert technologique) The process of converting scientific findings from research laboratories into useful products by the commercial sector.

Telocentric (French: télocentrique) Centromere located at the very end of a chromosome.

Telomere (French: télomère) The ends of chromosomes. These specialized structures are involved in the replication and stability of linear DNA molecules. See DNA replication.

Template, RNA or DNA (French: matrice d'ARN ou ADN) The base sequences found in nucleic acids that serve as the basis for the synthesis of complementary strrands of DNA or RNA.

Teratogen (French: tératogène) Substance or agent that can induce congenital malformations by action on the embryo. Thalidomide is a teratogenic agent.

Teratoma (French: tératome) Tumoral formation of embryonic origin made of tissues of endoderm,ectoderm and mesoderm origin in which we may find nervous cells, hair, teeth that have no connection with the surrounding tissues.The teratoma are sometime called twins with an imperfect development.

Terminal transferase (French: transférase terminale) Enzyme that has the capacity to add a desoxy ribonucleotide in the 3'OH of a DNA strand.

Terminator (French: terminateur) DNA sequence that initiates the end of the transcription.

Tetraploidy (French: tétraploïdie) Cellular content is made of 4n or 92 chromosomes.

Tétrasomy (French: tétrasomie) Presence of two chromosomes in addition to te normal pair of homologue chromosomes. Example tetrasomy X in a 48 chromosome cell.

Tetravalent (French: tétravalent) When two chromosomes are translocated in the first meiotic division; there is then a four chromosome formation called tetravalent instead of two bivalent homologue chromosomes.

Thenar (French: éminence thénar) Dermal ridges between the 5th finger and the wrist.

Thymine,T (French: thymine,T) A nitrogenous base, one member of the base pair A- T<adenine- thymine>.

Totipotent, cells (French: cellules totipotentes) Cells of the very early embryo that have the capacity to differentiation into the placenta the embryo and all post-embryonic tissues and organs. No stem cell line to date has been able to show these properties.

Trait (French: trait) Element transmitted by heredity. Recessive character present in a heterozygote form, like for instance the thalassemia trait.

Trans control (French: contrôle en trans) Regulation of genetic expression that manifests through an intermediate diffusible factor.

Transcriptase reverse (French: transcriptase inverse) Enzymatic complex present in RNA viruses assuring the DNA synthesis from RNA.

Transcription (French: transcription) The synthesis of an RNA copy from a sequence of DNA or gene; the first step in gene expression.

Transcription initiation site (French: site d'initiation de la transcription) Exact site of the initiation of transcription.

Transcription unit (French: unité de transcription) Region of the genome located between an initiation site and a site of termination of the transcription by the DNA polymerase.

Transcriptional readthrough (French: lecture transcriptionnelle) Uninterrupted DNA transcription.

Transducing phage (French: phage transducteur) A virus particle that accidentally contains a very small portion of its host chromosome, used in . They can program other strains of bacteria to manufacture proteins they normally cannot make.

Transductor phage (French: phage transducteur) Phage that has the property to transfer part of a genome from the host cell to another.

Transfection (French: transfection) A gene that upon transfection converts a previously immortalized cell to the malignant phenotype.

Transfer RNA (French: ARN de transfert, ARNt) A class of RNA having structures with triplet nucleotide sequences that are complementary to the triplet nucleotide coding sequences of mRNA. The role of tRNAs in protein synthesis is to bond with amino acids and transfer them to the ribosomes, where proteins are assembled according to the genetic code carried by mRNA.

Transformation (French: transformation) A process by which the genetic material carried by an individual cell is altered by incorporation of exogenous DNA into its genome.

Transgene (French: transgène) Gene introduced in the genome of an organism by genetics engineering.

Transgenesis (French: transgénèse) Operation that consists in producing transgenic organisms.

Transgenic, animal (French: animal transgénique) In reference to a living organism which foreign DNA has been introduced in all or the majority of his cells. The estrange gene can be transmitted to descents. Animal conceived to bear a gene from another animal.

Transient expression (French: expression transitoire) Expression of a gene trecently introduced into a cell and not yet integrated into the genome.

Transition (French: transition) A mutation in which either purine is substituted for the other, A for G or G for A or one pyrimidine is substituted for the other C for T or T for C.

Traduction (French: traduction) The process of forming a specific protein having its own amino acid sequence determined by the codons of the messenger RNA. The ribosome and transfer RNA are necessary for this process.

Translation error, reading mistake (French: erreur de traduction) Incorrect placement of one or

more amino acid residues in a polypeptide chain during genetic translation.

Translational control (French: contrôle de la traduction) The regulation of gene expression at the level of genetic translation.

Translational readtrough (French: translecture traductionnelle) Translation of a mRNA at ahead of the normal termination codon.

Translocation (French: translocation) The transfer of a segment of one chromosome to another chromosome.

Translocation, balanced (French: translocation équilibrée) The transfer of a segment of one chromosome to another without loss or addition of chromosomal material.

Translocation, insertional (French: translocation insertionnelle) The transfer of a segment of one chromosome to another chromosome by insertion of the segment following two breaks in the chromosome followed by reunion of the chromosome segments involved.

Translocation, reciprocal (French: translocation réciproque) Exchange of chromosome pieces between two non homologous chromosomes.

Translocation robertsonian (French: translocation robertsonienne) Translocation of two acrocentric chromosomes by fusion at or near the centromere, with loss of the short arms.

Transposase (French: transposase) Enzyme coded by a gene carried by a transposable involved in the transposition.

Transposition (French: transposition) Change of position of a DNA fragment in the genome.

Transposon (French: gène sauteur, transposon) DNA fragment susceptible to move from one location to another in the genome.

Transversion (French: transversion) A mutation in which either purine is substituted for either pyrimidine or vice versa.

Tricuspid atresia (French: atrésie tricuspidienne) Atresia of the heart pulmonary valve between the right cardiac auricle and the right ventricle.

Triplet, nonsense, see : nonsense codon.

Triploidy (French: diandrie, triploïdie, digynie) A cell with three copies of each chromosoome or an individual made up of such cells.

Triradius (French: triradius) Point of origin of dermal ridges in 3 directions.

Trisomy (French: trisomie) The state of having three representatives of a given chromosome instead of the usual pair, as in trisomy 21 or Down syndrome.

Tropism cellular (French: cellular tropism) Property of a virus to infect a cell type preferentially.

True hermaphrodyte (French: hermaphrodisme vrai) Hermaphrodyte individual who has gonadal tissue of both sexes.

Trunctus arteriosus (French: trunctus arteriosus) Congenital heart anomaly caracterized by the presence of only one arterial structure giving rise to aortic and pulmonary branches.

Tumor suppressor gene (French: Gène suppresseur de tumeur) A normal gene involved in the regulation of cell growth. Recessive mutations can lead to tumor development, as in the retinoblastoma gene or the p53 gene.

Tumor transformation (French: transformation tumorale) Conversion of eucaryotic cells into a stase of unrestrained growth in culture resembling or identical with the tumorigenic condition.

Twinning, see gemellity.

U

Ubiquitar molecule (French: molecule ubiquitaire) Molecule that is present in several tissues of the body.

Unequal crossing over (French: malségrégation) Crossing over between similar DNA sequences that are misaligned, resulting in sequences with deletion or duplication of DNA segments. A cause of a number of genetic variants.

Unequal division, see: unequal crossing over.

Uniparental disomy (French: disomie uniparentale) Presence in a diploid cell of two homologue chromosomes inherited from the same parent. The most common mechanism is probably the correction of a trisomic cell.

UPD, see uniparental disomy.

Uracil, U (French : uracil, U) A nitrogenous base normally found in RNA but not DNA. Uracil is capable of forming of forming a base pair with Adenine.

V

Vector (French : vecteur) See cloning vector.

Vector, expression (French: vecteur d'expression) Vector that has a region that allows the insertion of a gene coding sequence between the signals essential for its expression.

Ventricular septal defect (French: communication inter-ventriculaire) Congenital heart defect. Persistance of a communication between the two ventricles.

Villi, see: chorionic villi sampling.

Virulent phage, see lytic phage.

Virus (French: virus) A noncellular biological entity that can reproduce only within a host cell. Viruses consist of nucleic acid covered by protein; some animal viruses are also surrounded by membrane. Inside the infected cell, the virus uses the synthetic capability of the host to produce progeny virus.

Virus defective (French: virus défectif) Mutant virus that can reproduce only in the presence of an assistant virus.

Virus, helper (French: virus assistant) A defective gene that uses single–stranded RNA as its genetic material. It is only able to replicate when a helper virus is present in the same host cell.

Virus VONC (French: virus VONC) Gene originating from a proto-oncogene and subsequently recuperated by a virus.

VLSI (French: VLSI) Very large-scale integration allowing over 100,000 transistors on a chip.

VONC, see virus Vonc.

W

Wild (French: sauvage) The normal allele of a rare mutant gene, sometimes symbolized by +.

Williams, syndrome (French: syndrome de Williams)

Whorl (French: tourbillon) Dermatoglyphic pattern observed on the finger tips.

X

Xenograft (French: xénogreffes) Living cells, tissues and organs used in the xenografts Transfer of cells, tissues and organs between two organisms that belong to different species.

Xenotransplantation (French: xénotransplantation) Transfer of cells, tissues or living organs from living animals to humans for medical purposes.

X fragile, see Fragile X.

Y

YAC (French: YAC) See yeast artificial chromosome.

Yeast artificial chromosome,YAC (French: chromosome artificiel de levure,YAC) A vector used to clone DNA fragments (up to 400 kb); it is constructed from the telomeric, centromeric, and replication origin sequences needed for replication in yeast cells. Also see: cloning vector, cosmid.

Z

Zinc finger proteins (French: proteines à doigt de zinc) Transcription activator proteins, containing finger like structures containing zinc atoms.

Zoonoses (French: zoonoses) Animal diseases that can be transmitted to humans in living conditions. Animals and humans can be infected by zoonoses. Examples: rabies, brucellosis, mad cow disease or Creutzfeldt-Jakob disease in humans

Zygote (French: zygote) A fertilized egg formed as the result of the union of male and female sex cell –sperm and egg-.

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